

Canine Genetic Health Certificate™

Call Name:	Oakland	Laboratory #:	428037
Registered Name:	-	Registration #:	-
Breed:	Border Collie	Certificate Date:	Dec. 18, 2023
Sex:	Female		
DOB:	Sept. 2023		

This canine's DNA showed the following genotype(s):

Disease	Gene	Genotype	Interpretation
Collie Eye Anomaly	<i>NHEJ1</i>	WT/WT	Normal (Clear)
Degenerative Myelopathy (Common Variant)	<i>SOD1</i>	WT/WT	Normal (Clear)
Dental Hypomineralization	<i>FAM20C</i>	WT/WT	Normal (Clear)
Exercise-Induced Collapse	<i>DNM1</i>	WT/WT	Normal (Clear)
Glaucoma (Border Collie Type)	<i>OLFML3</i>	WT/WT	Normal (Clear)
Hyperuricosuria	<i>SLC2A9</i>	WT/WT	Normal (Clear)
Intestinal Cobalamin Malabsorption (Border Collie Type)	<i>CUBN</i>	WT/WT	Normal (Clear)
Multidrug Resistance 1	<i>ABCB1</i>	WT/WT	Normal (Clear)
Myotonia Congenita (Australian Cattle Dog Type)	<i>CLCN1</i>	WT/WT	Normal (Clear)
Neuronal Ceroid Lipofuscinosis 5 (Herding Dog Type)	<i>CLN5</i>	WT/WT	Normal (Clear)
Sensory Neuropathy (Border Collie Type)	<i>FAM134B</i>	WT/WT	Normal (Clear)
Trapped Neutrophil Syndrome	<i>VPS13B</i>	WT/M	Carrier

WT, wild type (normal); M, mutant; Y, Y chromosome (male)

Paw Print Genetics® performed the testing on the dog listed on this certificate. See the Laboratory Report for interpretation and recommendations based on these findings. The genes/diseases reported here were selected by the client. Normal results do not exclude inherited mutations not tested in these or other genes that may cause medical problems or may be passed on to offspring. The results included in this report relate only to the items tested using the sample provided. These tests were developed and their performance determined by Paw Print Genetics. This laboratory has established and verified the test(s) accuracy and precision with >99.9% sensitivity and specificity. The presence of mosaicism may not be detected by this test. Non-paternity may lead to unexpected results. This is not a breed identification test. Because all tests performed are DNA-based, rare genomic variations may interfere with the performance of some tests producing false results. If you think these results are in error, please contact the laboratory immediately for further evaluation. In the event of a valid dispute of results claim, Paw Print Genetics will do its best to resolve such a claim to the customer's satisfaction. If no resolution is possible after investigation by Paw Print Genetics with the cooperation of the customer, the extent of the customer's sole remedy is a refund of the fee paid. In no event shall Paw Print Genetics be liable for indirect, consequential or incidental damages of any kind. Any claim must be asserted within 60 days of the report of the test results. Genetic counseling is available at Paw Print Genetics.

Coat Color and Trait Certificate

Call Name:	Oakland	Laboratory #:	428037
Registered Name:	-	Registration #:	-
Breed:	Border Collie	Certificate Date:	Dec. 18, 2023
Sex:	Female		
DOB:	Sept. 2023		

This canine's DNA showed the following genotype(s):

Coat Color/Trait Test	Gene	Genotype	Interpretation
A Locus (Agouti)	<i>ASIP</i>	a^t/a^t	Tricolor, black and tan
A^S Locus (Saddle Tan)	<i>RALY</i>	N/N	No saddle tan/creeping tan
B Locus (Brown)	<i>TYRP1</i>	B/B	Black coat, nose and foot pads (does not carry brown)
Co Locus (Cocoa, French Bulldog Type)	<i>HPS3</i>	CO/CO	Black coat, nose and foot pads (does not carry cocoa)
D Locus (Dilute)	<i>MLPH</i>	d^1/d^1	Dilute (carries two copies of dilute)
E Locus	<i>MC1R</i>	E^m/E	Melanistic Mask - Carrier (Black)
H Locus (Harlequin, Great Dane Type)	<i>PSMB7</i>	h/h	No harlequin
I Locus (Intensity)	<i>MFSD12</i>	I/I	Normal intensity
K Locus (Dominant Black)	<i>CBD103</i>	k^y/k^y	Agouti expression allowed
M Locus (Merle)	<i>PMEL</i>	NA	Non merle
R Locus (Roan/Ticked)	<i>USH2A</i>	r/r	No roan or ticking
S Locus (White Spotting, Parti, or Piebald)	<i>MITF</i>	S/S	No white spotting, flash, parti, or piebald
SD Locus (Shedding)	<i>MC5R</i>	SD/SD	High shedding

Interpretation:

This dog carries two copies of a^t which results in tan points and can also present as a black and tan or tricolor coat color. However, this dog's coat color is also dependent on the E, K, and B genes. The tan point coat color is only expressed if the dog is also E/E or E/e at the E locus and k^y/k^y at the K locus. This dog will pass on a^t to 100% of its offspring.

This dog carries two copies of the **N** allele, which is not associated with a saddle tan coat color. This dog's coat color is also dependent on the E, A, and K genes, among others. This dog will pass **N** to 100% of its offspring.

This dog does not carry any copies of the b^a , b^c , b^d , b^e , b^h , and b^s mutations and has a B locus genotype of B/B. Thus, this dog typically will have a black coat, nose, and foot pads. However, this dog's coat color is dependent on the genotypes of many other genes. This dog will pass one copy of B to 100% of its offspring and cannot produce b/b dogs.

This dog does not carry any copies of the co (cocoa) mutation and has a Co Locus genotype of **CO/CO**. Thus, this dog typically will have a black coat, nose, and foot pads. However, this dog's coat color is dependent on the genotypes of many other genes including the B Locus (Brown). This dog will pass one copy of **CO** to 100% of its offspring and cannot produce co/co (cocoa) dogs.

This dog carries two copies of the d¹ mutation and has a D locus genotype of d/d which results in the dilution or lightening of the pigments that produce the dog's coat color. This dog will pass one copy of d¹ to 100% of its offspring. This dog can produce d/d offspring if bred to a dog that is also a carrier of a d mutation (D/d or d/d).

This dog carries one copy of **E^m** and one copy of **E** which allows for the production of black pigment with a melanistic mask on the muzzle of the dog. However, a melanistic mask may be unrecognizable on a dog with a dark coat color. This dog's coat color is also dependent on the K, A, and B genes. This dog will pass **E^m** on to 50% of its offspring and **E** to 50% of its offspring, which can produce black coat color if inherited with another copy of E, e^A, e¹, e², or e³.

This dog carries two copies of **h** and will not have a harlequin coat color. The dog will pass on **h** to 100% of its offspring.

This dog does not carry a copy of the i mutation and has an I locus genotype of **I/I** which does not result in the lightening of the light, phaeomelanin pigments that produce the dog's coat color in an e/e dog. This dog will pass one copy of **I** to 100% of its offspring and cannot produce i/i dogs.

This dog carries two copies of **k^Y** which allows for the expression of the agouti gene (A locus) which can result in a variety of coat colors including sable/fawn, tricolor, tan points, black or brown. However, this dog's coat color is dependent on its genotypes at the E, A and B genes. This dog will pass on **k^Y** to 100% of its offspring.

This dog carries two copies of **m**, the non-merle, wild-type allele of the *PMEL* gene, and, therefore, does not have a merle coat color/pattern. This dog will pass on one copy of the **m** allele to 100% of its offspring.

This dog does not carry a copy of R or R^{Ti} and has a genotype of r/r which does not result in roan or ticking in the white areas of the coat. This dog will pass one copy of r to 100% of its offspring.

This dog carries two copies of **S** which results in a solid coat with no white spotting, flash, parti, or piebald coat color. This dog will pass on one copy of **S** to 100% of its offspring.

This dog carries two copies of **SD** which has been associated with higher shedding. However, the overall degree of shedding for this dog is dependent on the combination of this dog's genotypes at the SD and IC loci. This dog will pass **SD** on to 100% of its offspring.

Paw Print Genetics® has genetic counseling available to you at no additional charge to answer any questions about these test results, their implications and potential outcomes in breeding this dog.

Paw Print Genetics® performed the testing on the dog listed on this certificate. The genes/traits reported here were selected by the client. Normal results do not exclude inherited mutations not tested in these or other genes that may cause variation in traits, medical problems or may be passed on to offspring. The results included in this report relate only to the items tested using the sample provided. These tests were developed and their performance determined by Paw Print Genetics. This laboratory has established and verified the test(s)' accuracy and precision with >99.9% sensitivity and specificity. The presence of mosaicism may not be detected by this test. Non-paternity may lead to unexpected results. This is not a breed identification test. Because all tests performed are DNA-based, rare genomic variations may interfere with the performance of some tests producing false results. If you think any results are in error, please contact the laboratory immediately for further evaluation. In the event of a valid dispute of results claim, Paw Print Genetics will do its best to resolve such a claim to the customer's satisfaction. If no resolution is possible after investigation by Paw Print Genetics with the cooperation of the customer, the extent of the customer's sole remedy is a refund of the fee paid. In no event shall Paw Print Genetics be liable for indirect, consequential or incidental damages of any kind. Any claim must be asserted within 60 days of the report of the test results.

Oakland

Breed: Border Collie
Birth date: 2023-09-27

Test date: 2024-03-20
ID kit: DKCGFJX

Oakland's Profile

Pet information

Registered name Oakland	Sex F
Owner reported breed Border Collie	Date of birth 2023-09-27

Genetic Diversity

Oakland's Percentage of Heterozygosity
38%

Health summary

- At Risk** 0 conditions
- Carrier** 1 condition
 - Trapped Neutrophil Syndrome
- Clear** 271 conditions

Oakland

Breed: Border Collie
Birth date: 2023-09-27

Test date: 2024-03-20
ID kit: DKCGFJX

Genetic Diversity

Heterozygosity

Oakland's Percentage of Heterozygosity

38%

Oakland's genome analysis shows an average level of genetic heterozygosity when compared with other Border Collies.

Typical Range for Border Collies

32% - 39%

Breed: Border Collie
Birth date: 2023-09-27Test date: 2024-03-20
ID kit: DKCGFJX

Health conditions known in the breed

Trapped Neutrophil Syndrome

Gene	Risk Variant	Copies	Inheritance	Result
VPS13B	Deletion	1	AR	Carrier

Information about the genetic condition

Clinical signs of TNS include an exceptional susceptibility to infections secondary to the low number of circulating neutrophils in the blood stream. Affected dogs also tend to suffer from chronic inflammatory conditions such as arthritis. Clinical signs are usually observed by 6 to 12 weeks of age and can include a smaller overall size as well as a ferret-like face due to abnormal craniofacial development leading to a narrowed, elongated skull shape. For some affected dogs, clinical signs can be mild and go unnoticed until adulthood. Nevertheless, TNS is a severe disease and affected dogs have a shorter life expectancy.

Breeder recommendation

This disease is autosomal recessive meaning that two copies of the mutation are needed for disease signs to occur. A carrier dog with one copy of the TNS mutation can be safely bred with a clear dog with no copies of the TNS mutation. About half of the puppies will have one copy (carriers) and half will have no copies of the TNS mutation. Puppies in a litter which is expected to contain carriers should be tested prior to breeding. Carrier to carrier matings are not advised as the resulting litter may contain affected puppies. Please note: It is possible that disease signs similar to the ones caused by the TNS mutation could develop due to a different genetic or clinical cause.

Collie Eye Anomaly (CEA)

Gene	Risk Variant	Copies	Inheritance	Result
NHEJ1	Deletion	0	AR	Clear

Information about the genetic condition

Collie Eye Anomaly is primarily characterized by choroidal hypoplasia, leading to an underdeveloped vascular supply to the retina, and is especially visible temporal to the optic nerve. CEA lesions may be present in both eyes or asymmetric in nature. CEA-associated choroidal hypoplasia is non-progressive and usually does not cause visual deficits on its own. However, CEA has a range of clinical expressions. Vision impairment is more likely in dogs with the "extended CEA phenotype," which may include optic nerve head colobomas, retinal detachment or intraocular hemorrhage secondary to coloboma(s) in severely affected dogs. Optic nerve head colobomas appear as excavations of the optic disc surface. Diagnosis of CEA lesions should be completed before 10 weeks of age, as retinal pigmentation can mask choroidal hypoplasia as the puppies grow, a phenomenon termed "go normal" by breeders. Research is ongoing to determine what additional genetic factors may be present that influence the range of severity seen in dogs with CEA.

Breeder recommendation

This disorder is autosomal recessive, meaning two copies of the variant are needed for a dog to be at an elevated risk for being diagnosed with the condition. A carrier dog with one copy of the Collie Eye Anomaly variant can be safely bred with a clear dog with no copies of the Collie Eye Anomaly variant. About half of the puppies will have one copy (carriers) and half will have no copies of the variant. Furthermore, a dog with two copies of the CEA variant can be safely bred with a clear dog. The resulting puppies will all be carriers. Puppies in a litter which is expected to contain carriers should be tested prior to breeding. Carrier to carrier matings are not advised as the resulting litter may contain affected puppies. Please note: Recent research has suggested that additional genetic risk factors likely exist in some breeds that resemble or contribute to CEA risk, especially the more severe disorder expression. It is possible that disorder signs similar to the ones associated with this CEA variant could develop due to a different genetic or clinical cause.

Breed: Border Collie
Birth date: 2023-09-27

Test date: 2024-03-20
ID kit: DKCGFJX

Health conditions known in the breed

Dental Hypomineralization	Gene	Risk Variant	Copies	Inheritance	Result
	FAM20C	C>T	0	AR	Clear

Information about the genetic condition

Clinical signs include brownish dental discoloration and abnormal wear of teeth. As the teeth wear, the biting surfaces of the teeth darkens, become dark brown in color; the enamel layer may also show a light brown discoloration and appear dull. The disorder causes severe tooth wear leading to pulp exposure, chronic inflammation of the pulp, and pulpal necrosis. Histologically, dentin of affected dogs has an abnormal structure and the enamel can be slightly hypoplastic.

Breeder recommendation

This disease is autosomal recessive meaning that two copies of the mutation are needed for disease signs to be shown. A carrier dog with one copy of the Dental Hypomineralization mutation can be safely bred with a clear dog with no copies of the Dental Hypomineralization mutation. About half of the puppies will have one copy (carriers) and half will have no copies of the Dental Hypomineralization mutation. A dog with two copies of the Dental Hypomineralization mutation can be safely bred with a clear dog. The resulting puppies will all be carriers. Puppies in a litter which is expected to contain carriers should be tested prior to breeding. Carrier to carrier matings are not advised as the resulting litter may contain affected puppies. Please note: It is possible that disease signs similar to the ones caused by the Dental Hypomineralization mutation could develop due to a different genetic or clinical cause.

Early Adult Onset Deafness For Border Collies only (Linkage test)	Gene	Risk Variant	Copies	Inheritance	Result
	Intergenic	Insertion	0	AR	Clear

Information about the genetic condition

Gradual hearing loss affecting both ears is observed usually between the ages of 5 to 7 years. Please note that this test is specifically for the Border Collie breed and is a predictive linkage test rather than a test for the true causal variant. Not all dogs with two copies of the linked marker will go on to show signs of hearing loss.

Breeder recommendation

This disease is autosomal recessive meaning that two copies of the mutation are needed for disease signs to develop. A carrier dog with one copy of the Deafness mutation can be safely bred with a clear dog with no copies of the Deafness mutation. About half of the puppies will have one copy (carriers) and half will have no copies of the Deafness mutation. Puppies in a litter which is expected to contain carriers should be tested prior to breeding. The carrier rate of the risk variant is up to 35% in the Border Collie population, highlighting the importance of keeping healthy carriers in the breeding program by breeding them to dogs tested "Clear" (zero copies) of the risk variant. Please note: It is possible that disease signs similar to the ones caused by the Deafness mutation could develop due to a different genetic or clinical cause.

Breed: Border Collie
Birth date: 2023-09-27

Test date: 2024-03-20
ID kit: DKCGFJX

Health conditions known in the breed

Hereditary Calcium Oxalate Urolithiasis, Type 1	Gene	Risk Variant	Copies	Inheritance	Result
	Confidential	-	0	AR	Clear

Information about the genetic condition

Hereditary Calcium Oxalate Urolithiasis, Type 1 is a disorder that is associated with increased risk of urinary calcium oxalate stone formation. Affected dogs will demonstrate clinical signs consistent with urolithiasis. This may range from being asymptomatic to hematuria (bloody urine), dysuria (painful urination), stranguria (straining to pass urine) and pollakiuria (frequent urination). Dogs with urinary stones are also more susceptible to urinary tract infections. And, due to the presence of the stones, affected dogs are at risk of urinary obstruction occurring at the renal pelvis, ureters, or urethra. Blockage of the urinary tract is a life-threatening condition that requires immediate intervention. While the average age of diagnosis is 3 years old, dogs affected by CaOx1 have the potential to develop urinary stones as puppies. And recurrent stone formation is common for affected dogs. There is evidence to suggest the clinical signs are more common in males than in females.

Breeder recommendation

This disorder is autosomal recessive, meaning two copies of the variant are needed for a dog to be at an elevated risk for being diagnosed with the condition. A carrier dog with one copy of the Hereditary Calcium Oxalate Urolithiasis, Type 1 variant can be safely bred with a clear dog with no copies of the Hereditary Calcium Oxalate Urolithiasis, Type 1 variant. About half of the puppies will have one copy (carriers) and half will have no copies of the variant. Furthermore, a dog with two copies of the Hereditary Calcium Oxalate Urolithiasis, Type 1 variant can be safely bred with a clear dog. The resulting puppies will all be carriers. Puppies in a litter which is expected to contain carriers should be tested prior to breeding. Carrier to carrier matings are not advised as the resulting litter may contain affected puppies. Please note: It is possible that disorder signs similar to the ones associated with this CaOx1 variant could develop due to a different genetic or clinical cause.

Hyperuricosuria	Gene	Risk Variant	Copies	Inheritance	Result
	SLC2A9	G>T	0	AR	Clear

Information about the genetic condition

HUU predisposes affected dogs to the formation of urate stones. Clinical signs of urolithiasis include hematuria, pain while urinating, and blockage of the urinary tract. Patients with urinary stones are more susceptible to urinary tract infections. Blockage of the urinary tract is a life-threatening condition that requires immediate veterinary care. In Dalmatians, the clinical signs are more common in males than in females. As many as 34% of all male Dalmatians are diagnosed with urate stones.

Breeder recommendation

This disease is autosomal recessive meaning that two copies of the mutation are needed for disease signs to occur. A carrier dog with one copy of the HUU mutation can be safely bred with a clear dog with no copies of the HUU mutation. About half of the puppies will have one copy (carriers) and half will have no copies of the HUU mutation. A dog with two copies of the HUU mutation can be safely bred with a clear dog. The resulting puppies will all be carriers. Puppies in a litter which is expected to contain carriers should be tested prior to breeding. In some breeds, such as the Dalmatian, the frequency of the disease mutation is very high. Carriers and dogs with two copies of the disease mutation (genetically affected dogs) should be used for breeding purposes, with the aim of gradually reducing the frequency of the mutant gene within the breed population. Where possible, matings should be avoided that would result in litters that could contain dogs with two copies of the disease mutation, such as a mating between two dogs with two copies of the HUU mutation or between a dog with one copy and a dog with two copies of the HUU mutation. Please note: It is possible that disease signs similar to the ones caused by the HUU mutation could develop due to a different genetic or clinical cause.

Breed: Border Collie
Birth date: 2023-09-27

Test date: 2024-03-20
ID kit: DKCGFJX

Health conditions known in the breed

Intestinal Cobalamin Malabsorption (Discovered in the Border Collie)	Gene	Risk Variant	Copies	Inheritance	Result
	CUBN	Deletion	0	AR	Clear

Information about the genetic condition

Initial signs of intestinal cobalamin malabsorption can be seen in puppies 6 to 12 weeks of age, when cobalamin store become depleted. Puppies with IGS suffer from weakness and loss of appetite and fail to grow normally Bloodwork shows anemia, neutropenia, and low cobalamin concentrations. High levels of homocysteine and methylmalonic acid can also be observed in the blood. Proteinuria is typically present.

Breeder recommendation

This disease is autosomal recessive meaning that two copies of the mutation are needed for disease signs to occur. A carrier dog with one copy of the ICM mutation can be safely bred with a clear dog with no copies of the ICM mutation. About half of the puppies will have one copy (carriers) and half will have no copies of the ICM mutation. A dog with two copies of the ICM mutation can be safely bred with a clear dog. The resulting puppies will all be carriers. Puppies in a litter which is expected to contain carriers should be tested prior to breeding. Carrier to carrier matings are not advised as the resulting litter may contain affected puppies. Please note: It is possible that disease signs similar to the ones caused by the ICM mutation could develop due to a different genetic or clinical cause.

MDR1 Medication Sensitivity	Gene	Risk Variant	Copies	Inheritance	Result
	MDR1/ABCB1	Deletion	0	AD	Clear

Information about the genetic condition

Dogs with this variant are asymptomatic until exposed to a medication that uses the drug transport pump rendered defective by the mutation in the MDR1 (also called ABCB1) gene. Medications known to use this P-glycoprotein pump are macrocyclic lactones (antiparasitic drugs), loperamide (antidiarrheal), erythromycin (antibiotic), acepromazine (tranquilizer), butorphanol (opioid), certain drugs used in cancer treatment (vincristine, vinblastine, and doxorubicin), and others. When these medications are administered, they accumulate in the brain which results in adverse reactions. Typical symptoms include tremors, loss of balance, seizures, obtundation, excessive salivation, dilated pupils, and bradycardia. If untreated, the condition may lead to respiratory arrest, coma or death. Because dogs with 1 copy of the variant will have some P-glycoprotein function, the most severe cases tend to occur in dogs that have 2 copies of the variant and, therefore, lack any functional P-glycoprotein pumps. However, the disorder can still be very severe in dogs that have only one copy of the mutation.

Breeder recommendation

This disorder is autosomal dominant meaning that only one copy of the variant is needed for associated signs to occur. For some breeds where the MDR1 mutation frequency is particularly high, breeders may consider mating pairs using dogs that have one or two copies of the MDR1 variant to maintain genetic diversity within their breed. It is important that resulting puppies be tested for the MDR1 variant to ensure safe future medical treatment. If a dog with one copy of the MDR1 variant is bred with a clear dog with no copies of the MDR1 variant, about half of the puppies will have one copy and half will have no copies of the MDR1 variant. If a dog with two copies of the MDR1 variant is bred with a clear dog, the resulting puppies will all have one copy of the variant. Please note: It is possible that clinical signs similar to the ones caused by the MDR1 variant could develop due to a different genetic or clinical cause.

Breed: Border Collie
Birth date: 2023-09-27

Test date: 2024-03-20
ID kit: DKCGFJX

Health conditions known in the breed

Neuronal Ceroid Lipofuscinosis 5 (Discovered in the Border Collie)	Gene	Risk Variant	Copies	Inheritance	Result
	CLN5	C>T	0	AR	Clear

Information about the genetic condition

Neuronal ceroid lipofuscinoses (NCLs) are a group of inherited progressive neurodegenerative lysosomal storage disorders. NCLs are characterized by excessive accumulation of lipofuscin and ceroid lipopigments in the central nervous system and other tissues. The age of onset for dogs affected with Neuronal Ceroid Lipofuscinosis 5 (NCL5) can vary significantly, with some showing initial signs at 1 to 2 years of age while others show later in life. Similarly, severity of clinical signs can vary between affected individuals. Typical signs of NCL5 include vision impairment, epileptic seizures, ataxia (uncoordinated movements), and behavioral changes, such as hyperactivity and aggression. Some affected dogs can show air biting, likely secondary to hallucinations. Due to the progressive nature of NCL5, the average prognosis is considered poor for affected dogs. And the average life expectancy is less than 2.5 years.

Breeder recommendation

This disorder is autosomal recessive, meaning two copies of the variant are needed for a dog to be at an elevated risk for being diagnosed with the condition. A carrier dog with one copy of the Neuronal Ceroid Lipofuscinosis 5 (Discovered in the Border Collie) variant can be safely bred with a clear dog with no copies of the Neuronal Ceroid Lipofuscinosis 5 (Discovered in the Border Collie) variant. About half of the puppies will have one copy (carriers) and half will have no copies of the variant. Puppies in a litter which is expected to contain carriers should be tested prior to breeding. Carrier to carrier matings are not advised as the resulting litter may contain affected puppies. Please note: It is possible that disorder signs similar to the ones associated with this NCL5 variant could develop due to a different genetic or clinical cause.

Sensory Neuropathy	Gene	Risk Variant	Copies	Inheritance	Result
	FAM134B	Insertion	0	AR	Clear

Information about the genetic condition

Clinical signs are detectable in puppies from two to seven months of age. Clinical signs include incoordination of gait (ataxia), knuckling of the paws, hyperextension of the limbs, and self-mutilation of the limbs. The hind legs are usually most severely affected. Loss of sensation is progressive and affects all limbs. Urinary incontinence and regurgitation can occur in the later stages of the disorder.

Breeder recommendation

This disease is autosomal recessive meaning that two copies of the mutation are needed for disease signs to develop. A carrier dog with one copy of the Sensory Neuropathy mutation can be safely bred with a clear dog with no copies of the Sensory Neuropathy mutation. About half of the puppies will have one copy (carriers) and half will have no copies of the Sensory Neuropathy mutation. Puppies in a litter which is expected to contain carriers should be tested prior to breeding. Carrier to carrier matings are not advised as the resulting litter may contain affected puppies. Please note: It is possible that disease signs similar to the ones caused by the Sensory Neuropathy mutation could develop due to a different genetic or clinical cause.

Breed: Border Collie
Birth date: 2023-09-27

Test date: 2024-03-20
ID kit: DKCGFJX

Traits

Coat Color

	Gene	Variant	Copies	Result
Fawn	ASIP	ay	0	No effect
Recessive Black	ASIP	a	0	No effect
Tan Points Two copies, or occasionally one copy, of this variant may result in a black and tan coat color pattern.	ASIP	at	2	Tan points possible
Dominant Black	CBD103	KB	0	No effect
Mask One or two copies of the Mask mutation will result in the presence of a dark facial mask covering the muzzle. This mask can cover only the very front of the muzzle, or can extend down to the chest and front legs. Mask can be hidden by other trait variants.	MC1R	Em	1	Dark Muzzle possible
Recessive Red (e1)	MC1R	e1	0	No effect
Recessive Red (e2)	MC1R	e2	0	No effect
Recessive Red (e3)	MC1R	e3	0	No effect
Sable (Discovered in the Cocker Spaniel)	MC1R	eH	0	No effect
Widow's Peak (Discovered in Ancient dogs)	MC1R	eA	0	No effect
Widow's Peak (Discovered in the Afghan Hound and Saluki)	MC1R	eG	0	No effect

Color Modification

	Gene	Variant	Copies	Result
Cocoa (Discovered in the French Bulldog)	HPS3	co	0	No effect
Red Intensity	MFS12	i	0	No effect

Breed: Border Collie
Birth date: 2023-09-27

Test date: 2024-03-20
ID kit: DKCGFJX

Color Modification

	Gene	Variant	Copies	Result
Dilution (d1) Linkage test To show coat color dilution, a dog must inherit two copies of a dilution variant, one from each parent. This can either be two copies of a particular variant, such as this one (d1) or two of any combination of dilution variants. This variant (d1) is the most common dilution variant in dogs. The test for d1 is a linkage test, that measures markers close to the d1 variant to determine the most likely d1 genotype. The test is 99.2% accurate based on a set of over 3000 breed and mixed breed dogs with a known d1 genotype.	MLPH	d ¹	2	Dilution likely
Dilution (d2)	MLPH	d ²	0	No effect
Dilution (d3)	MLPH	d ³	0	No effect
Chocolate (basd)	TYRP1	b ^{asd}	0	No effect
Chocolate (bc)	TYRP1	b ^c	0	No effect
Chocolate (bd)	TYRP1	b ^d	0	No effect
Chocolate (be)	TYRP1	b ^e	0	No effect
Chocolate (bh)	TYRP1	b ^h	0	No effect
Chocolate (bs)	TYRP1	b ^s	0	No effect

Coat Patterns

	Gene	Variant	Copies	Result
Piebald	MITF	s ^p	0	No effect
Merle	PMEL	M	0	No effect
Harlequin	PSMB7	H	0	No effect
Saddle Tan	RALY	-	0	No effect
Roan Linkage Test	USH2A	T ^r	0	No effect

Breed: Border Collie
Birth date: 2023-09-27

Test date: 2024-03-20
ID kit: DKCGFJX

Coat Length and Curl

	Gene	Variant	Copies	Result
Long Hair (lh1) To show a long coat, a dog must inherit two copies of a Long Hair variant, one from each parent. This can either be two copies of a particular variant, such as this one (lh1) or two of any combination of long hair variants. However, there are other variants suspected to influence coat length.	FGF5	lh ¹	2	Long coat
Long Hair (lh2)	FGF5	lh ²	0	No effect
Long Hair (lh3)	FGF5	lh ³	0	No effect
Long Hair (lh4)	FGF5	lh ⁴	0	No effect
Long Hair (lh5)	FGF5	lh ⁵	0	No effect
Curly Coat	KRT71	C	0	No effect

Hairlessness

	Gene	Variant	Copies	Result
Hairlessness (Discovered in the Chinese Crested Dog) Linkage test	FOXI3	H ^{rcc}	0	No effect
Hairlessness (Discovered in the American Hairless Terrier)	SGK3	h ^{raht}	0	No effect
Hairlessness (Discovered in the Scottish Deerhound)	SKG3	h ^{rsd}	0	No effect

Shedding

	Gene	Variant	Copies	Result
Reduced Shedding	MC5R	sd	0	Seasonal shedder

Breed: Border Collie
Birth date: 2023-09-27Test date: 2024-03-20
ID kit: DKCGFJX

More Coat Traits

	Gene	Variant	Copies	Result
Hair Ridge	FGF3, FGF4, FGF19, ORAOV1	R	0	No effect
Furnishings	RSPO2	F	0	No effect
Albino	SLC45A2	ca1	0	No effect

Head Shape

	Gene	Variant	Copies	Result
Short Snout (BMP3 variant)	BMP3	-	0	No effect
Short Snout (SMOC2 variant)	SMOC2	-	0	No effect

Eye Color

	Gene	Variant	Copies	Result
Blue Eyes (Discovered in the Siberian Husky)	ALX4	-	0	No effect

Ears

	Gene	Variant	Copies	Result
Floppy Ears	MSRB3	-	0	Pricked ears more likely

Extra Toes

	Gene	Variant	Copies	Result
Hind Dewclaws (Discovered in Asian breeds)	LMBR1	DC-1	0	No effect

Breed: Border Collie
Birth date: 2023-09-27

Test date: 2024-03-20
ID kit: DKCGFJX

Extra Toes

	Gene	Variant	Copies	Result
Hind Dewclaws (Discovered in Western breeds) One or two copies of this Hind Dewclaws variant may result in your dog having hind dewclaws. Around half of the dogs with one copy of this variant will have hind dewclaws, and it is possible for the dewclaws to be just on one leg. With two copies the trait is more likely to be expressed and could be more pronounced.	LMBR1	DC-2	1	Hind dewclaws possible

More Body Features

	Gene	Variant	Copies	Result
Back Muscle and Bulk	ACSL4	-	0	No effect
High Altitude Adaptation	EPAS1	-	0	No effect
Short Legs (Chondrodysplasia, CDPA)	FGF4	-	0	No effect
Short Legs (Chondrodystrophy, CDDY)	FGF4	-	0	No effect
Short Tail	T-box	T	0	Full tail length likely

Breed: Border Collie
Birth date: 2023-09-27Test date: 2024-03-20
ID kit: DKCGFJX

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
2,8-dihydroxyadenine (DHA) Urolithiasis	APRT	G>A	0	AR	Clear
Acral Mutilation Syndrome	GDNF	C>T	0	AR	Clear
Acute Respiratory Distress Syndrome	ANLN	C>T	0	AR	Clear
Alaskan Husky Encephalopathy	SLC19A3	G>A	0	AR	Clear
Alexander Disease	GFAP	G>A	0	AR	Clear
Amelogenesis Imperfecta (Discovered in the Italian Greyhound)	ENAM	Deletion	0	AR	Clear
Amelogenesis Imperfecta (Discovered in the Lancashire Heeler)	Confidential	-	0	AR	Clear
Amelogenesis Imperfecta (Discovered in the Parson Russell Terrier)	ENAM	C>T	0	AR	Clear
Bandera's Neonatal Ataxia	GRM1	Insertion	0	AR	Clear
Benign Familial Juvenile Epilepsy	LGI2	A>T	0	AR	Clear
Bernard-Soulier Syndrome (Discovered in the Cocker Spaniel)	GP9	Deletion	0	AR	Clear
Canine Congenital Stationary Night Blindness (Discovered in the Beagle)	LRIT3	Deletion	0	AR	Clear
Canine Leukocyte Adhesion Deficiency (CLAD), type III	FERMT3	Insertion	0	AR	Clear
Canine Multifocal Retinopathy 1	BEST1	C>T	0	AR	Clear
Canine Multifocal Retinopathy 2	BEST1	G>A	0	AR	Clear
Canine Multifocal Retinopathy 3	BEST1	Deletion	0	AR	Clear
Canine Multiple Systems Degeneration (Discovered in the Chinese Crested Dog)	SERAC1	Deletion	0	AR	Clear
Canine Scott Syndrome	ANO6	G>A	0	AR	Clear
Cardiomyopathy and Juvenile Mortality (Discovered in the Belgian Shepherd)	YARS2	G>A	0	AR	Clear
Centronuclear Myopathy (Discovered in the Great Dane)	BIN1	A>G	0	AR	Clear

Breed: Border Collie
Birth date: 2023-09-27

Test date: 2024-03-20
ID kit: DKCGFJX

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Centronuclear Myopathy (Discovered in the Labrador Retriever)	PTPLA	Insertion	0	AR	Clear
Cerebellar Ataxia	RAB24	A>C	0	AR	Clear
Cerebellar Cortical Degeneration	SNX14	C>T	0	AR	Clear
Cerebellar Hypoplasia	VLDLR	Deletion	0	AR	Clear
Cerebral Dysfunction	SLC6A3	G>A	0	AR	Clear
Chondrodysplasia (Discovered in Norwegian Elkhound and Karelian Bear Dog)	ITGA10	C>T	0	AR	Clear
Chondrodystrophy (CDDY) and Intervertebral Disc Disease (IVDD) Risk	FGF4 retrogene	Insertion	0	AD	Clear
Cleft Lip & Palate with Syndactyly	ADAMTS20	Deletion	0	AR	Clear
Cleft Palate	DLX6	C>A	0	AR	Clear
CNS Atrophy with Cerebellar Ataxia (Discovered in the Belgian Shepherd)	SEPP1	Deletion	0	AR	Clear
Coat Color Dilution and Neurological Defects (Discovered in the Miniature Dachshund)	MYO5A	Insertion	0	AR	Clear
Complement 3 Deficiency	C3	Deletion	0	AR	Clear
Cone Degeneration (Discovered in the Alaskan Malamute)	CNGB3	Deletion	0	AR	Clear
Cone Degeneration (Discovered in the German Shepherd Dog)	CNGA3	C>T	0	AR	Clear
Cone Degeneration (Discovered in the German Shorthaired Pointer)	CNGB3	G>A	0	AR	Clear
Cone-Rod Dystrophy	NPHP4	Deletion	0	AR	Clear
Cone-Rod Dystrophy 1	PDE6B	Deletion	0	AR	Clear
Cone-Rod Dystrophy 2	IQCB1	Insertion	0	AR	Clear
Congenital Cornification (Discovered in the Labrador Retriever)	NSDHL	Deletion	0	XD	Clear

Breed: Border Collie
Birth date: 2023-09-27Test date: 2024-03-20
ID kit: DKCGFJX

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Congenital Dys hormonogenic Hypothyroidism with Goiter (Discovered in the Shih Tzu)	SLC5A5	G>A	0	AR	Clear
Congenital Eye Malformations (Discovered in the Golden Retriever)	SIX6	C>T	0	AD	Clear
Congenital Hypothyroidism (Discovered in the Tenterfield Terrier)	TPO	C>T	0	AR	Clear
Congenital Hypothyroidism (Discovered in the Toy Fox and Rat Terrier)	TPO	C>T	0	AR	Clear
Congenital Muscular Dystrophy (Discovered in the Italian Greyhound)	LAMA2	G>A	0	AR	Clear
Congenital Muscular Dystrophy (Discovered in the Staffordshire Bull Terrier)	LAMA2	Deletion	0	AR	Clear
Congenital Myasthenic Syndrome (Discovered in the Golden Retriever)	COLQ	G>A	0	AR	Clear
Congenital Myasthenic Syndrome (Discovered in the Heideterrier)	CHRNE	Insertion	0	AR	Clear
Congenital Myasthenic Syndrome (Discovered in the Jack Russell Terrier)	CHRNE	Insertion	0	AR	Clear
Congenital Myasthenic Syndrome (Discovered in the Labrador Retriever)	COLQ	T>C	0	AR	Clear
Congenital Myasthenic Syndrome (Discovered in the Old Danish Pointer)	CHAT	G>A	0	AR	Clear
Congenital Stationary Night Blindness (CSNB)	RPE65	A>T	0	AR	Clear
Craniomandibular Osteopathy (Discovered in Scottish Terrier breeds)	SLC37A2	C>T	0	AD	Clear
Craniomandibular Osteopathy (Discovered in the Australian Terrier)	COL1A1	C>T	0	AD	Clear
Craniomandibular Osteopathy (Discovered in the Basset Hound)	SLC37A2	C>T	0	AD	Clear
Craniomandibular Osteopathy (Discovered in the Weimaraner)	SLC35D1	Deletion	0	AD	Clear
Cystic Renal Dysplasia and Hepatic Fibrosis	INPP5E	G>A	0	AR	Clear

Breed: Border Collie
Birth date: 2023-09-27Test date: 2024-03-20
ID kit: DKCGFJX

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Cystinuria Type I-A	SLC3A1	C>T	0	AR	Clear
Cystinuria Type II-A	SLC3A1	Deletion	0	AD	Clear
Darier Disease (Discovered in the Irish Terrier)	ATP2A2	Insertion	0	AD	Clear
Deafness and Vestibular Dysfunction (DINGS1), (Discovered in Doberman Pinscher)	PTPRQ	Insertion	0	AR	Clear
Deafness and Vestibular Dysfunction (DINGS2), (Discovered in Doberman Pinscher)	MYO7A	G>A	0	AR	Clear
Degenerative Myelopathy	SOD1	G>A	0	AR	Clear
Demyelinating Neuropathy	SBF2	G>T	0	AR	Clear
Dental-Skeletal-Retinal Anomaly (Discovered in the Cane Corso)	MIA3	Deletion	0	AR	Clear
Dilated Cardiomyopathy (Discovered in the Schnauzer)	RBM20	Deletion	0	AR	Clear
Disproportionate Dwarfism (Discovered in the Dogo Argentino)	PRKG2	C>A	0	AR	Clear
Dominant Progressive Retinal Atrophy	RHO	C>G	0	AD	Clear
Dystrophic Epidermolysis Bullosa (Discovered in the Basset Hound)	COL7A1	Insertion	0	AR	Clear
Dystrophic Epidermolysis Bullosa (Discovered in the Central Asian Ovcharka)	COL7A1	C>T	0	AR	Clear
Dystrophic Epidermolysis Bullosa (Discovered in the Golden Retriever)	COL7A1	C>T	0	AR	Clear
Early Retinal Degeneration (Discovered in the Norwegian Elkhound)	STK38L	Insertion	0	AR	Clear
Early-Onset Adult Deafness (Discovered in the Rhodesian Ridgeback)	EPS8L2	Deletion	0	AR	Clear
Early-Onset Progressive Polyneuropathy (Discovered in the Alaskan Malamute)	NDRG1	G>T	0	AR	Clear
Early-Onset Progressive Polyneuropathy (Discovered in the Greyhound)	NDRG1	Deletion	0	AR	Clear

Breed: Border Collie
Birth date: 2023-09-27Test date: 2024-03-20
ID kit: DKCGFJX

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Early-Onset Progressive Retinal Atrophy (Discovered in the Portuguese Water Dog)	CCDC66	Insertion	0	AR	Clear
Early-Onset Progressive Retinal Atrophy, (Discovered in the Spanish Water Dog)	PDE6B	Deletion	0	AR	Clear
Ehlers-Danlos Syndrome (Discovered in mixed breed)	COL5A1	G>A	0	AD	Clear
Ehlers-Danlos Syndrome (Discovered in the Labrador Retriever)	COL5A1	Deletion	0	AD	Clear
Epidermolytic Hyperkeratosis	KRT10	G>T	0	AR	Clear
Episodic Falling Syndrome	BCAN	Insertion	0	AR	Clear
Exercise-Induced Collapse	DNM1	G>T	0	AR	Clear
Factor VII Deficiency	F7	G>A	0	AR	Clear
Factor XI Deficiency	FXI	Insertion	0	AD	Clear
Familial Nephropathy (Discovered in the English Cocker Spaniel)	COL4A4	A>T	0	AR	Clear
Familial Nephropathy (Discovered in the English Springer Spaniel)	COL4A4	C>T	0	AR	Clear
Fanconi Syndrome	FAN1	Deletion	0	AR	Clear
Fetal Onset Neuroaxonal Dystrophy	MFN2	G>C	0	AR	Clear
Focal Non-Epidermolytic Palmoplantar Keratoderma	KRT16	G>C	0	AR	Clear
Generalized Progressive Retinal Atrophy (Discovered in the Schapendoes)	CCDC66	Insertion	0	AR	Clear
Glanzmann Thrombasthenia Type I (Discovered in Great Pyrenees)	ITGA2B	C>G	0	AR	Clear
Glanzmann Thrombasthenia Type I (Discovered in mixed breed dogs)	ITGA2B	C>T	0	AR	Clear
Globoid Cell Leukodystrophy (Discovered in Terriers)	GALC	A>C	0	AR	Clear
Globoid Cell Leukodystrophy (Discovered in the Irish Setter)	GALC	A>T	0	AR	Clear

Breed: Border Collie
Birth date: 2023-09-27

Test date: 2024-03-20
ID kit: DKCGFJX

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Glycogen Storage Disease Type Ia (Discovered in the German Pinscher)	G6PC	Insertion	0	AR	Clear
Glycogen Storage Disease Type Ia (Discovered in the Maltese)	G6PC	G>C	0	AR	Clear
Glycogen Storage Disease Type IIIa, (GSD IIIa)	AGL	Deletion	0	AR	Clear
GM1 Gangliosidosis (Discovered in the Portuguese Water Dog)	GLB1	G>A	0	AR	Clear
GM1 Gangliosidosis (Discovered in the Shiba)	GLB1	Deletion	0	AR	Clear
GM2 Gangliosidosis (Discovered in the Japanese Chin)	HEXA	G>A	0	AR	Clear
GM2 Gangliosidosis (Discovered in the Toy Poodle)	HEXB	Deletion	0	AR	Clear
Hemophilia A (Discovered in Old English Sheepdog)	FVIII	C>T	0	XR	Clear
Hemophilia A (Discovered in the Boxer)	FVIII	C>G	0	XR	Clear
Hemophilia A (Discovered in the German Shepherd Dog - Variant 1)	FVIII	G>A	0	XR	Clear
Hemophilia A (Discovered in the German Shepherd Dog - Variant 2)	FVIII	G>A	0	XR	Clear
Hemophilia A (Discovered in the Havanese)	FVIII	Insertion	0	XR	Clear
Hemophilia A (Discovered in the Labrador Retriever)	Confidential	-	0	XR	Clear
Hemophilia B	FIX	G>A	0	XR	Clear
Hemophilia B (Discovered in the Airedale Terrier)	FIX	Insertion	0	XR	Clear
Hemophilia B (Discovered in the Lhasa Apso)	FIX	Deletion	0	XR	Clear
Hereditary Ataxia (Discovered in the Belgian Malinois)	SLC12A6	Insertion	0	AR	Clear
Hereditary Ataxia (Discovered in the Norwegian Buhund)	KCNIP4	T>C	0	AR	Clear
Hereditary Elliptocytosis	SPTB	C>T	0	AD	Clear
Hereditary Footpad Hyperkeratosis	FAM83G	G>C	0	AR	Clear

Breed: Border Collie
Birth date: 2023-09-27Test date: 2024-03-20
ID kit: DKCGFJX

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Hereditary Nasal Parakeratosis (Discovered in the Greyhound)	SUV39H2	Deletion	0	AR	Clear
Hereditary Nasal Parakeratosis (Discovered in the Labrador Retriever)	SUV39H2	A>C	0	AR	Clear
Hereditary Vitamin D-Resistant Rickets Type II	VDR	Deletion	0	AR	Clear
Hypocatalasia	CAT	G>A	0	AR	Clear
Hypomyelination	FNIP2	Deletion	0	AR	Clear
Hypophosphatasia	Confidential	-	0	AR	Clear
Ichthyosis (Discovered in the American Bulldog)	NIPAL4	Deletion	0	AR	Clear
Ichthyosis (Discovered in the Great Dane)	SLC27A4	G>A	0	AR	Clear
Ichthyosis Type 2 (Discovered in the Golden Retriever)	ABHD5	Deletion	0	AR	Clear
Inflammatory Myopathy (Discovered in the Dutch Shepherd Dog)	SLC25A12	A>G	0	AR	Clear
Inflammatory Pulmonary Disease (Discovered in the Rough Collie)	AKNA	Deletion	0	AR	Clear
Intestinal Cobalamin Malabsorption (Discovered in the Beagle)	CUBN	Deletion	0	AR	Clear
Intestinal Cobalamin Malabsorption (Discovered in the Komondor)	CUBN	G>A	0	AR	Clear
Intestinal Lipid Malabsorption (Discovered in the Australian Kelpie)	ACSL5	Deletion	0	AR	Clear
Junctional Epidermolysis Bullosa (Discovered in the Australian Cattle Dog Mix)	LAMA3	T>A	0	AR	Clear
Junctional Epidermolysis Bullosa (Discovered in the Australian Shepherd)	LAMB3	A>G	0	AR	Clear
Juvenile Cataract (Discovered in the Wirehaired Pointing Griffon)	FYCO1	Deletion	0	AR	Clear
Juvenile Dilated Cardiomyopathy (Discovered in the Toy Manchester Terrier)	ABCC9	G>A	0	AR	Clear

Breed: Border Collie
Birth date: 2023-09-27Test date: 2024-03-20
ID kit: DKCGFJX

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Juvenile Encephalopathy (Discovered in the Parson Russell Terrier)	Confidential	-	0	AR	Clear
Juvenile Laryngeal Paralysis and Polyneuropathy	RAB3GAP1	Deletion	0	AR	Clear
Juvenile Myoclonic Epilepsy	DIRAS1	Deletion	0	AR	Clear
L-2-Hydroxyglutaric aciduria (Discovered in the Staffordshire Bull Terrier)	L2HGDH	T>C	0	AR	Clear
L-2-Hydroxyglutaric Aciduria (Discovered in the West Highland White Terrier)	Confidential	-	0	AR	Clear
Lafora Disease (Linkage test)	NHLRC1	Insertion	0	AR	Clear
Lagotto Storage Disease	ATG4D	G>A	0	AR	Clear
Lamellar Ichthyosis	TGM1	Insertion	0	AR	Clear
Laryngeal Paralysis (Discovered in the Bull Terrier and Miniature Bull Terrier)	RAPGEF6	Insertion	0	AR	Clear
Leigh-like Subacute Necrotizing Encephalopathy (Discovered in the Yorkshire Terrier)	SLC19A3	Insertion	0	AR	Clear
Lethal Acrodermatitis (Discovered in the Bull Terrier)	MKLN1	A>C	0	AR	Clear
Leukodystrophy (Discovered in the Standard Schnauzer)	TSEN54	C>T	0	AR	Clear
Ligneous Membranitis	PLG	T>A	0	AR	Clear
Limb-girdle Muscular Dystrophy (Discovered in the Boston Terrier) Variant 1	SGCD	Deletion	0	AR	Clear
Limb-girdle Muscular Dystrophy, Type L3 (Discovered in the Miniature Dachshund)	SGCA	G>A	0	AR	Clear
Lung Developmental Disease (Discovered in the Airedale Terrier)	LAMP3	C>T	0	AR	Clear
Macrothrombocytopenia (Discovered in Norfolk and Cairn Terrier)	TUBB1	G>A	0	AR	Clear
May-Hegglin Anomaly	MYH9	G>A	0	AD	Clear
Microphthalmia (Discovered in the Soft-Coated Wheaten Terrier)	RBP4	Deletion	0	AR	Clear

Breed: Border Collie
Birth date: 2023-09-27

Test date: 2024-03-20
ID kit: DKCGFJX

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Mucopolysaccharidosis Type IIIA (Discovered in the Dachshund)	SGSH	C>A	0	AR	Clear
Mucopolysaccharidosis Type IIIA (Discovered in the New Zealand Huntaway)	SGSH	Insertion	0	AR	Clear
Mucopolysaccharidosis Type VII (Discovered in the Brazilian Terrier)	GUSB	C>T	0	AR	Clear
Mucopolysaccharidosis Type VII (Discovered in the German Shepherd Dog)	GUSB	G>A	0	AR	Clear
Mucopolysaccharidosis VI (Discovered in the Miniature Pinscher)	ARSB	G>A	0	AR	Clear
Muscular Dystrophy (Discovered in the Cavalier King Charles Spaniel)	Dystrophin	G>T	0	XR	Clear
Muscular Dystrophy (Discovered in the Golden Retriever)	Dystrophin	A>G	0	XR	Clear
Muscular Dystrophy (Discovered in the Landseer)	COL6A1	G>T	0	AR	Clear
Muscular Dystrophy (Discovered in the Norfolk Terrier)	Dystrophin	Deletion	0	XR	Clear
Muscular Dystrophy-Dystroglycanopathy (Discovered in the Labrador Retriever)	LARGE	C>T	0	AR	Clear
Muscular Hypertrophy (Double Muscling)	MSTN	T>A	0	AR	Clear
Musladin-Lueke Syndrome	ADAMTSL2	C>T	0	AR	Clear
Myeloperoxidase Deficiency	MOP	C>T	0	AR	Clear
Myotonia Congenita (Discovered in Australian Cattle Dog)	CLCN1	Insertion	0	AR	Clear
Myotonia Congenita (Discovered in the Labrador Retriever)	CLCN1	T>A	0	AR	Clear
Myotonia Congenita (Discovered in the Miniature Schnauzer)	CLCN1	C>T	0	AR	Clear
Myotubular Myopathy	MTM1	A>C	0	XR	Clear
Narcolepsy (Discovered in the Dachshund)	HCRTR2	G>A	0	AR	Clear
Narcolepsy (Discovered in the Labrador Retriever)	HCRTR2	G>A	0	AR	Clear

Breed: Border Collie
Birth date: 2023-09-27Test date: 2024-03-20
ID kit: DKCGFJX

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Nemaline Myopathy	NEB	C>A	0	AR	Clear
Neonatal Cerebellar Cortical Degeneration	SPTBN2	Deletion	0	AR	Clear
Neonatal Encephalopathy with Seizures	ATF2	T>G	0	AR	Clear
Neuroaxonal Dystrophy (Discovered in Spanish Water Dog)	TECPR2	C>T	0	AR	Clear
Neuroaxonal Dystrophy (Discovered in the Papillon)	PLA2G6	G>A	0	AR	Clear
Neuroaxonal Dystrophy (Discovered in the Rottweiler)	VPS11	A>G	0	AR	Clear
Neuronal Ceroid Lipofuscinosis 1	PPT1	Insertion	0	AR	Clear
Neuronal Ceroid Lipofuscinosis 12 (Discovered in the Australian Cattle Dog)	ATP13A2	C>T	0	AR	Clear
Neuronal Ceroid Lipofuscinosis 5 (Discovered in the Golden Retriever)	CLN5	Deletion	0	AR	Clear
Neuronal Ceroid Lipofuscinosis 7	MFSD8	Deletion	0	AR	Clear
Neuronal Ceroid Lipofuscinosis 8 (Discovered in the Alpine Dachsbracke)	CLN8	Deletion	0	AR	Clear
Neuronal Ceroid Lipofuscinosis 8 (Discovered in the Australian Shepherd)	CLN8	G>A	0	AR	Clear
Neuronal Ceroid Lipofuscinosis 8 (Discovered in the English Setter)	CLN8	T>C	0	AR	Clear
Neuronal Ceroid Lipofuscinosis 8 (Discovered in the Saluki)	CLN8	Insertion	0	AR	Clear
Obesity risk (POMC)	POMC	Deletion	0	AD	Clear
Osteochondrodysplasia	SLC13A1	Deletion	0	AR	Clear
Osteochondromatosis (Discovered in the American Staffordshire Terrier)	EXT2	C>A	0	AR	Clear
Osteogenesis Imperfecta (Discovered in the Beagle)	COL1A2	C>T	0	AD	Clear
Osteogenesis Imperfecta (Discovered in the Dachshund)	SERPINH1	T>C	0	AR	Clear

Breed: Border Collie
Birth date: 2023-09-27Test date: 2024-03-20
ID kit: DKCGFJX

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
P2RY12-associated Bleeding Disorder	P2RY12	Deletion	0	AR	Clear
Palmoplantar Hyperkeratosis (Discovered in the Rottweiler)	DSG1	Deletion	0	AR	Clear
Paroxysmal Dyskinesia	PIGN	C>T	0	AR	Clear
Persistent Müllerian Duct Syndrome	AMHR2	C>T	0	AR	Clear
Phosphofruktokinase Deficiency	PFKM	G>A	0	AR	Clear
Pituitary Dwarfism (Discovered in the Karelian Bear Dog)	POU1F1	C>A	0	AR	Clear
Polycystic Kidney Disease	PKD1	G>A	0	AD	Clear
Prekallikrein Deficiency	KLKB1	T>A	0	AR	Clear
Primary Ciliary Dyskinesia	CCDC39	C>T	0	AR	Clear
Primary Ciliary Dyskinesia (Discovered in the Alaskan Malamute)	NME5	Deletion	0	AR	Clear
Primary Lens Luxation	ADAMTS17	G>A	0	AR	Clear
Primary Open Angle Glaucoma (Discovered in Basset Fauve de Bretagne)	ADAMTS17	G>A	0	AR	Clear
Primary Open Angle Glaucoma (Discovered in Petit Basset Griffon Vendeen)	ADAMTS17	Insertion	0	AR	Clear
Primary Open Angle Glaucoma and Lens Luxation (Discovered in Chinese Shar-Pei)	ADAMTS17	Deletion	0	AR	Clear
Progressive Early-Onset Cerebellar Ataxia	SEL1L	T>C	0	AR	Clear
Progressive Retinal Atrophy (Discovered in the Basenji)	SAG	T>C	0	AR	Clear
Progressive Retinal Atrophy (Discovered in the Golden Retriever - GR-PRA 2 variant)	TTC8	Deletion	0	AR	Clear
Progressive Retinal Atrophy (Discovered in the Golden Retriever - GR-PRA1 variant)	SLC4A3	Insertion	0	AR	Clear
Progressive Retinal Atrophy (Discovered in the Lapponian Herder)	IFT122	C>T	0	AR	Clear

Breed: Border Collie
Birth date: 2023-09-27Test date: 2024-03-20
ID kit: DKCGFJX

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Progressive Retinal Atrophy (Discovered in the Lhasa Apso)	IMPG2	Insertion	0	AR	Clear
Progressive Retinal Atrophy (Discovered in the Miniature Long Haired Dachshund)	RPGRIP1	Insertion	0	AR	Clear
Progressive Retinal Atrophy (Discovered in the Papillon and Phalène)	CNGB1	Deletion	0	AR	Clear
Progressive Retinal Atrophy (Discovered in the Shetland Sheepdog - BBS2 variant)	Confidential	-	0	AR	Clear
Progressive Retinal Atrophy (Discovered in the Shetland Sheepdog - CNGA1 variant)	CNGA1	Deletion	0	AR	Clear
Progressive Retinal Atrophy (Discovered in the Swedish Vallhund)	MERTK	Insertion	0	AR	Clear
Progressive Retinal Atrophy 1 (Discovered in the Italian Greyhound)	Confidential	-	0	AR	Clear
Progressive Retinal Atrophy Type III	FAM161A	Insertion	0	AR	Clear
Progressive Rod Cone Degeneration (prcd-PRA)	PRCD	G>A	0	AR	Clear
Protein Losing Nephropathy	NPHS1	G>A	0	AR	Clear
Pyruvate Dehydrogenase Phosphatase 1 Deficiency	PDP1	C>T	0	AR	Clear
Pyruvate Kinase Deficiency (Discovered in the Basenji)	PKLR	Deletion	0	AR	Clear
Pyruvate Kinase Deficiency (Discovered in the Beagle)	PKLR	G>A	0	AR	Clear
Pyruvate Kinase Deficiency (Discovered in the Pug)	PKLR	T>C	0	AR	Clear
Pyruvate Kinase Deficiency (Discovered in the West Highland White Terrier)	PKLR	Insertion	0	AR	Clear
QT Syndrome	KCNQ1	C>A	0	AD	Clear
Renal Cystadenocarcinoma and Nodular Dermatofibrosis	FLCN	A>G	0	AD	Clear
Rod-Cone Dysplasia 1	PDE6B	G>A	0	AR	Clear
Rod-Cone Dysplasia 1a	PDE6B	Insertion	0	AR	Clear

Breed: Border Collie
Birth date: 2023-09-27Test date: 2024-03-20
ID kit: DKCGFJX

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Rod-Cone Dysplasia 3	PDE6A	Deletion	0	AR	Clear
Sensorineural Deafness (Discovered in the Rottweiler)	LOXHD1	G>C	0	AR	Clear
Sensory Ataxic Neuropathy	tRNATyr	Deletion	0	MT	Clear
Severe Combined Immunodeficiency (Discovered in Frisian Water Dogs)	RAG1	G>T	0	AR	Clear
Severe Combined Immunodeficiency (Discovered in Russell Terriers)	PRKDC	G>T	0	AR	Clear
Shaking Puppy Syndrome (Discovered in the Border Terrier)	Confidential	-	0	AR	Clear
Skeletal Dysplasia 2	COL11A2	G>C	0	AR	Clear
Spinocerebellar Ataxia (Late-Onset Ataxia)	CAPN1	G>A	0	AR	Clear
Spinocerebellar Ataxia with Myokymia and/or Seizures	KCNJ10	C>G	0	AR	Clear
Spondylocostal Dysostosis	HES7	Deletion	0	AR	Clear
Spongy Degeneration with Cerebellar Ataxia (Discovered in Belgian Malinois - SDCA1)	KCNJ10	T>C	0	AR	Clear
Spongy Degeneration with Cerebellar Ataxia (Discovered in Belgian Malinois - SDCA2)	ATP1B2	Insertion	0	AR	Clear
Stargardt Disease (Discovered in the Labrador Retriever)	ABCA4	Insertion	0	AR	Clear
Startle Disease (Discovered in Irish Wolfhounds)	SLC6A5	G>T	0	AR	Clear
Startle Disease (Discovered in the Miniature American Shepherd)	Confidential	-	0	AR	Clear
Succinic Semialdehyde Dehydrogenase Deficiency (Discovered in the Saluki)	ALDH5A1	G>A	0	AR	Clear
Thrombopathia (Discovered in the Basset Hound)	RASGRP1	Deletion	0	AR	Clear
Thrombopathia (Discovered in the Eskimo Spitz)	RASGRP1	Insertion	0	AR	Clear
Van den Ende-Gupta Syndrome	SCARF2	Deletion	0	AR	Clear
von Willebrand's Disease, type 1	VWF	G>A	0	AD	Clear

Breed: Border Collie
Birth date: 2023-09-27Test date: 2024-03-20
ID kit: DKCGFJX

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
von Willebrand's Disease, type 2	VWF	T>G	0	AR	Clear
von Willebrand's Disease, type 3 (Discovered in the Kooiker Hound)	VWF	G>A	0	AR	Clear
von Willebrand's Disease, type 3 (Discovered in the Scottish Terrier)	VWF	Deletion	0	AR	Clear
von Willebrand's Disease, type 3 (Discovered in the Shetland Sheepdog)	VWF	Deletion	0	AR	Clear
X-Linked Ectodermal Dysplasia	EDA	G>A	0	XR	Clear
X-Linked Hereditary Nephropathy (Discovered in the Navasota Dog)	COL4A5	Deletion	0	XR	Clear
X-Linked Hereditary Nephropathy (Discovered in the Samoyed)	COL4A5	G>T	0	XR	Clear
X-Linked Myotubular Myopathy	MTM1	C>A	0	XR	Clear
X-Linked Progressive Retinal Atrophy 1	RPGR	Deletion	0	XR	Clear
X-Linked Progressive Retinal Atrophy 2	RPGR	Deletion	0	XR	Clear
X-Linked Severe Combined Immunodeficiency (Discovered in the Basset Hound)	IL2RG	Deletion	0	XR	Clear
X-Linked Severe Combined Immunodeficiency (Discovered in the Cardigan Welsh Corgi)	IL2RG	Insertion	0	XR	Clear
X-Linked Tremors	PLP1	A>C	0	XR	Clear
Xanthinuria (Discovered in a mixed breed dog)	Confidential	-	0	AR	Clear
Xanthinuria (Discovered in the Cavalier King Charles Spaniel)	Confidential	-	0	AR	Clear
Xanthinuria (Discovered in the Toy Manchester Terrier)	Confidential	-	0	AR	Clear

Breed: Border Collie
Birth date: 2023-09-27

Test date: 2024-03-20
ID kit: DKCGFJX

Glossary of genetic terms

Test result definitions

At Risk: Based on the disorder's mode of inheritance, the dog inherited a number of genetic variant(s) which increases the dog's risk of being diagnosed with the associated disorder.

Carrier: The dog inherited one copy of a genetic variant when two copies are usually necessary to increase the dog's risk of being diagnosed with the associated disorder. While carriers are usually not at risk of clinical expression of the disorder, carriers of some complex variants may be associated with a low risk of developing the disorder.

Clear: The dog did not inherit the genetic variant(s) associated with the disorder and will not be at elevated risk of being diagnosed with the disorder due to this genotype. However, similar clinical signs could develop from different genetic or clinical causes.

Inconclusive: An inconclusive result indicates a confident call could not be made based on the data for that genetic variant. Health testing is performed in replicates, and on occasion the outcomes do not agree. This may occur due to an unusual sequence of DNA in the region tested, multiple cell genotypes present due to chimerism or acquired mutations, or due to quality of the DNA sample.

Inheritance mode definitions

Autosomal Recessive (AR): For autosomal recessive disorders, dogs with two copies of the genetic variant are at risk of developing the associated disorder. Dogs with one copy of the variant are considered carriers and are usually not at risk of developing the disorder. However, carriers of some complex variants grouped in this category may be associated with a low risk of developing the disorder. Dogs with one or two copies may pass the disorder-associated variant to their puppies if bred.

Autosomal Dominant (AD): For autosomal dominant disorders, dogs with one or two copies of the genetic variant are at risk of developing the associated disorder. Inheriting two copies of the variant may increase the risk of development of the disorder or cause the condition to be more severe. These dogs may pass the disorder-associated variant to their puppies if bred.

X-linked Recessive (XR): For X-linked recessive disorders, the genetic variant is found on the X chromosome. Female dogs must inherit two copies of the variant to be at risk of developing the condition, whereas male dogs only need one copy to be at risk. Males and females with any copies of the variant may pass the disorder-associated variant to their puppies if bred.

X-linked Dominant (XD): For X-linked dominant disorders, the genetic variant is found on the X chromosome. Both male and female dogs with one copy of the variant are at risk of developing the disorder. Females inheriting two copies of the variant may be at higher risk or show a more severe form of the disorder than with one copy. Males and females with any copies of the variant may pass the disorder-associated variant to their puppies if bred.

Mitochondrial (MT): Unlike the two copies of genomic DNA held in the nucleus, there are thousands of mitochondria in each cell of the body, and each holds its own mitochondrial DNA (mtDNA). Mitochondria are called the "powerhouses" of the cell. For a dog to be at risk for a mitochondrial disorder, it must inherit a certain ratio of mtDNA with the associated variant compared to normal mtDNA. mtDNA is inherited only from the mother.