

# JAB DO RECANTO SÃO FRANCISCO

Breed: Border Collie

Birth date: 2020-04-12

Registration number: 2237

Test date: 2025-09-11

ID kit: DLWPKFM



## JAB DO RECANTO SÃO FRANCISCO's Profile

### Pet information

Registered name

JAB DO RECANTO SÃO FRANCISCO

Sex

M

Owner reported breed

Border Collie

Date of birth

2020-04-12

### Genetic Diversity

#### JAB DO RECANTO SÃO FRANCISCO's Percentage of Heterozygosity

33%

### Health summary

At Risk 0 conditions

Carrier 0 conditions

Clear 272 conditions

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## Genetic Diversity

### Heterozygosity

#### **JAB DO RECANTO SÃO FRANCISCO's Percentage of Heterozygosity**

33%

#### **Typical Range for Border Collies**

32% - 39%

JAB DO RECANTO SÃO FRANCISCO's genome analysis shows an average level of genetic heterozygosity when compared with other Border Collies.

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## Health conditions known in the breed

Collie Eye Anomaly (CEA)	Gene	Risk Variant	Copies	Inheritance	Result
	NHEJ1	Deletion	0	AR	<b>Clear</b>

### 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.

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

### 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.

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## Health conditions known in the breed

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

### 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.

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

### 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.

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## Health conditions known in the breed

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

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

Initial signs of intestinal cobalamin malabsorption can be seen in puppies 6 to 12 weeks of age, when cobalamin stores 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.

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## Health conditions known in the breed

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

### 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.

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

### 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.

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## Health conditions known in the breed

Sensory Neuropathy	Gene	Risk Variant	Copies	Inheritance	Result
	FAM134B	Insertion	0	AR	<b>Clear</b>

### 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.

Trapped Neutrophil Syndrome	Gene	Risk Variant	Copies	Inheritance	Result
	VPS13B	Deletion	0	AR	<b>Clear</b>

### 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.

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## Traits

### Coat Color

	Gene	Variant	Copies	Result
<b>Fawn</b>	ASIP	ay	0	No effect
<b>Recessive Black</b>	ASIP	a	0	No effect
<b>Tan Points</b>  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
<b>Dominant Black</b>  One or two copies of the dominant black will give a dog a black coat (depending on other variants), black eye rims, nose and pads. One copy may also give a tiger striped appearance, known as brindle patterning.	CBD103	K <sup>B</sup>	1	Black or brindle possible
<b>Mask</b>  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	E <sup>m</sup>	1	Dark Muzzle possible
<b>Recessive Red (e1)</b>	MC1R	e <sup>1</sup>	0	No effect
<b>Recessive Red (e2)</b>	MC1R	e <sup>2</sup>	0	No effect
<b>Recessive Red (e3)</b>	MC1R	e <sup>3</sup>	0	No effect
<b>Sable (Discovered in the Cocker Spaniel)</b>	MC1R	e <sup>H</sup>	0	No effect
<b>Widow's Peak (Discovered in Ancient dogs)</b>	MC1R	e <sup>A</sup>	0	No effect
<b>Widow's Peak (Discovered in the Afghan Hound and Saluki)</b>	MC1R	e <sup>G</sup>	0	No effect

### Color Modification

	Gene	Variant	Copies	Result
<b>Cocoa (Discovered in the French Bulldog)</b>	HPS3	co	0	No effect

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## Color Modification

	Gene	Variant	Copies	Result
<b>Red Intensity</b>  Dogs with two copies of the Red Intensity variant are more likely to show yellow, cream or white coat shades instead of deeper red shades. If the dog does not display solid red or red coat patterns, there will be no visible effect. Other genes, notably variants in the KITLG gene, are also thought to contribute to red pigment intensity variation, so some dogs may have yellow or buff colored coats.	MFSD12	i	1	No effect
<b>Dilution (d1) Linkage test</b>	MLPH	d <sup>1</sup>	0	No effect
<b>Dilution (d2)</b>	MLPH	d <sup>2</sup>	0	No effect
<b>Dilution (d3)</b>	MLPH	d <sup>3</sup>	0	No effect
<b>Chocolate (basd)</b>	TYRP1	b <sup>asd</sup>	0	No effect
<b>Chocolate (bc)</b>	TYRP1	b <sup>c</sup>	0	No effect
<b>Chocolate (bd)</b>	TYRP1	b <sup>d</sup>	0	No effect
<b>Chocolate (be)</b>	TYRP1	b <sup>e</sup>	0	No effect
<b>Chocolate (bh)</b>	TYRP1	b <sup>h</sup>	0	No effect
<b>Chocolate (bs)</b>	TYRP1	b <sup>s</sup>	0	No effect

## Coat Patterns

	Gene	Variant	Copies	Result
<b>Piebald</b>	MITF	s <sup>p</sup>	0	No effect
<b>Merle</b>	PMEL	M	0	No effect
<b>Harlequin</b>	PSMB7	H	0	No effect
<b>Saddle Tan</b>  One or two copies of the Saddle Tan variant are needed for the "saddle" to be seen. However the Tan Points variant must also be present. The Saddle Tan variant is actually considered to be the wild type, or default, variant.	RALY	-	1	Saddle possible

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## Coat Patterns

	Gene	Variant	Copies	Result
Roan Linkage Test	USH2A	TR <sup>r</sup>	0	No effect

## Coat Length and Curl

	Gene	Variant	Copies	Result
<b>Long Hair (lh1)</b>  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 <sup>1</sup>	1	Short coat likely, long coat possible
<b>Long Hair (lh2)</b>	FGF5	lh <sup>2</sup>	0	No effect
<b>Long Hair (lh3)</b>	FGF5	lh <sup>3</sup>	0	No effect
<b>Long Hair (lh4)</b>	FGF5	lh <sup>4</sup>	0	No effect
<b>Long Hair (lh5)</b>	FGF5	lh <sup>5</sup>	0	No effect
<b>Curly Coat</b>	KRT71	C	0	No effect

## Hairlessness

	Gene	Variant	Copies	Result
<b>Hairlessness (Discovered in the Chinese Crested Dog Linkage test)</b>	FOXI3	Hrc <sup>c</sup>	0	No effect
<b>Hairlessness (Discovered in the American Hairless Terrier)</b>	SGK3	hra <sup>ht</sup>	0	No effect
<b>Hairlessness (Discovered in the Scottish Deerhound)</b>	SGK3	hra <sup>sd</sup>	0	No effect

## Shedding

	Gene	Variant	Copies	Result
<b>Reduced Shedding</b>	MC5R	sd	0	Seasonal shedder

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## 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	c <sup>al</sup>	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
Hind Dewclaws (Discovered in Western breeds)	LMBR1	DC-2	0	No effect

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## 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

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## Other health conditions tested

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

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## Other health conditions tested

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

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## Other health conditions tested

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

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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

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## Other health conditions tested

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

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## Other health conditions tested

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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

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## Other health conditions tested

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

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## Other health conditions tested

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

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## Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
<b>Mucopolysaccharidosis Type IIIA (Discovered in the Dachshund)</b>	SGSH	C>A	0	AR	Clear
<b>Mucopolysaccharidosis Type IIIA (Discovered in the New Zealand Huntaway)</b>	SGSH	Insertion	0	AR	Clear
<b>Mucopolysaccharidosis Type VII (Discovered in the Brazilian Terrier)</b>	GUSB	C>T	0	AR	Clear
<b>Mucopolysaccharidosis Type VII (Discovered in the German Shepherd Dog)</b>	GUSB	G>A	0	AR	Clear
<b>Mucopolysaccharidosis VI (Discovered in the Miniature Pinscher)</b>	ARSB	G>A	0	AR	Clear
<b>Muscular Dystrophy (Discovered in the Cavalier King Charles Spaniel)</b>	Dystrophin	G>T	0	XR	Clear
<b>Muscular Dystrophy (Discovered in the Golden Retriever)</b>	Dystrophin	A>G	0	XR	Clear
<b>Muscular Dystrophy (Discovered in the Landseer)</b>	COL6A1	G>T	0	AR	Clear
<b>Muscular Dystrophy (Discovered in the Norfolk Terrier)</b>	Dystrophin	Deletion	0	XR	Clear
<b>Muscular Dystrophy-Dystroglycanopathy (Discovered in the Labrador Retriever)</b>	LARGE	C>T	0	AR	Clear
<b>Muscular Hypertrophy (Double Muscling)</b>	MSTN	T>A	0	AR	Clear
<b>Musladin-Lueke Syndrome</b>	ADAMTS2	C>T	0	AR	Clear
<b>Myeloperoxidase Deficiency</b>	MOP	C>T	0	AR	Clear
<b>Myotonia Congenita (Discovered in Australian Cattle Dog)</b>	CLCN1	Insertion	0	AR	Clear
<b>Myotonia Congenita (Discovered in the Labrador Retriever)</b>	CLCN1	T>A	0	AR	Clear
<b>Myotonia Congenita (Discovered in the Miniature Schnauzer)</b>	CLCN1	C>T	0	AR	Clear
<b>Myotubular Myopathy</b>	MTM1	A>C	0	XR	Clear
<b>Narcolepsy (Discovered in the Dachshund)</b>	HCRTR2	G>A	0	AR	Clear
<b>Narcolepsy (Discovered in the Labrador Retriever)</b>	HCRTR2	G>A	0	AR	Clear

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## Other health conditions tested

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

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

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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

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

# JAB DO RECANTO SÃO FRANCISCO

Breed: Border Collie  
Birth date: 2020-04-12

Registration number: 2237  
Test date: 2025-09-11  
ID kit: DLWPKFM



## Other health conditions tested

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

# JAB DO RECANTO SÃO FRANCISCO

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## 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.