

Shadow

Breed: Miniature Australian Shepherd Dog
Birth date: 2025-03-11

Test date: 2025-12-30
ID kit: DBZPKKPQMS

Shadow's Profile

Pet information

Registered name

Shadow

Sex

F

Owner reported breed

Miniature Australian Shepherd Dog

Date of birth

2025-03-11

Genetic Diversity

Shadow's Percentage of Heterozygosity

41%

Health summary

At Risk 0 conditions

Carrier 0 conditions

Clear 273 conditions

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Genetic Diversity

Heterozygosity

Shadow's Percentage of Heterozygosity

41%

Shadow's genome analysis shows an average level of genetic heterozygosity when compared with other Australian Shepherds.

Typical Range for Australian Shepherds

34% - 42%

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Health conditions known in the breed

Degenerative Myelopathy	Gene	Risk Variant	Copies	Inheritance	Result
	SOD1	G>A	0	AR	Clear

Information about the genetic condition

The onset of DM is usually in a dog's senior years. Affected dogs first begin by exhibiting muscle wasting, proprioceptive deficits, and knuckling of the hind feet. Though the condition is not painful, affected dogs will eventually require assistance walking. As the condition progresses, it moves up the spinal cord and the dog's neurologic deficits mirror the progress, losing fecal and urinary continence and eventually involving the front legs and the brainstem.

Breeder recommendation

Please note the DM mutation has only been associated with disease signs in some breeds, and this should be considered when making breeding decisions. 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 DM mutation can be safely bred with a clear dog with no copies of the DM mutation. About half of the puppies will have one copy (carriers) and half will have no copies of the DM mutation. A dog with two copies of the DM mutation can be safely bred with a clear dog. The resulting puppies will be all carriers with one copy of the DM mutation. Puppies in litters which are expected to contain carriers should be tested prior to breeding. Please note: It is possible that disease signs similar to the ones caused by the DM 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.

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Health conditions known in the breed

Progressive Rod Cone Degeneration (prcd-PRA)	Gene	Risk Variant	Copies	Inheritance	Result
	PRCD	G>A	0	AR	Clear

Information about the genetic condition

Clinical signs of PRCD are related to progressive loss of function of rod photoreceptors, followed by loss of function of cone photoreceptors. Typical signs of disease include hyper-reflective tapetum and attenuated blood vessels. Age of onset for this form of PRA is generally early adulthood, although exact age of onset may vary significantly among different breeds. The disorder is progressive, causing increasing levels of vision loss and eventual blindness.

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 prcd-PRA mutation can be safely bred with a clear dog with no copies of the prcd-PRA mutation. About half of the puppies will have one copy (carriers) and half will have no copies of the prcd-PRA mutation. A dog with two copies of the prcd-PRA 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 prcd-PRA mutation could develop due to a different genetic or clinical cause.

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Traits

Coat Color

	Gene	Variant	Copies	Result
Fawn	ASIP	ay	0	No effect
Recessive Black Two copies of the Recessive Black variant are needed for a black coat to be seen, and also an absence of other coat modifying variants such as chocolate and recessive red.	ASIP	a	1	Not black due to this variant
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	K ^B	0	No effect
Mask	MC1R	E ^m	0	No effect
Recessive Red (e1)	MC1R	e ¹	0	No effect
Recessive Red (e2)	MC1R	e ²	0	No effect
Recessive Red (e3)	MC1R	e ³	0	No effect
Sable (Discovered in the Cocker Spaniel)	MC1R	e ^H	0	No effect
Widow's Peak (Discovered in Ancient dogs)	MC1R	e ^A	0	No effect
Widow's Peak (Discovered in the Afghan Hound and Saluki)	MC1R	e ^G	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
Dilution (d1) Linkage test	MLPH	d ¹	0	No effect
Dilution (d2)	MLPH	d ²	0	No effect
Dilution (d3)	MLPH	d ³	0	No effect

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Color Modification

	Gene	Variant	Copies	Result
Chocolate (basd)	TYRP1	b ^{asd}	0	No effect
Chocolate (bc) To show chocolate coloration a dog must inherit two chocolate variants, one from each parent. This can either be two copies of a particular variant, such as this one ("bc"), or two of any combination of chocolate variants.	TYRP1	b ^c	1	Black features likely, chocolate possible
Chocolate (bd) To show chocolate coloration a dog must inherit two chocolate variants, one from each parent. This can either be two copies of a particular variant, such as this one ("bd"), or two of any combination of chocolate variants. This variant is unique in that it can occur on the same chromosome as another chocolate variant, where both variants are donated from one parent. If the other parent does not also donate a chocolate variant, the dog will still express black pigment, not chocolate.	TYRP1	b ^d	1	Black features likely, chocolate possible
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 Most dogs with one copy of the Merle variant will show Merle patterning. Most dogs with two copies will be mostly white, but in some cases will show Merle patterning. Some dogs with this variant will not show the Merle pattern. This is because the Merle variant can sometimes be shortened (known as cryptic or atypical Merle), and these forms do not have an effect on appearance.	PMEL	M	1	Merle possible
Harlequin	PSMB7	H	0	No effect
Saddle Tan	RALY	-	0	No effect

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Coat Patterns

	Gene	Variant	Copies	Result
Roan Linkage Test	USH2A	TR ^r	0	No effect

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	Hr ^{cc}	0	No effect
Hairlessness (Discovered in the American Hairless Terrier)	SGK3	hr ^{aht}	0	No effect
Hairlessness (Discovered in the Scottish Deerhound)	SKG3	hr ^{sd}	0	No effect

Shedding

	Gene	Variant	Copies	Result
Reduced Shedding	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	ca ^l	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) Dogs with one or two copies of the blue eye variant are likely to have one or two blue eyes.	ALX4	-	1	Blue eyes possible

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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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

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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
Collie Eye Anomaly (CEA)	NHEJ1	Deletion	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

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Congenital Cornification (Discovered in the Labrador Retriever)	NSDHL	Deletion	0	XD	Clear
Congenital Dyshormonogenic 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

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Cystic Renal Dysplasia and Hepatic Fibrosis	INPP5E	G>A	0	AR	Clear
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
Demyelinating Neuropathy	SBF2	G>T	0	AR	Clear
Dental Hypomineralization	FAM20C	C>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 Adult Onset Deafness For Border Collies only (Linkage test)	Intergenic	Insertion	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

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Early-Onset Progressive Polyneuropathy (Discovered in the Alaskan Malamute)	NDRG1	G>T	0	AR	Clear
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

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Globoid Cell Leukodystrophy (Discovered in the Irish Setter)	GALC	A>T	0	AR	Clear
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 Calcium Oxalate Urolithiasis, Type 1	Confidential	-	0	AR	Clear

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Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Hereditary Elliptocytosis	SPTB	C>T	0	AD	Clear
Hereditary Footpad Hyperkeratosis	FAM83G	G>C	0	AR	Clear
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
Hyperuricosuria	SLC2A9	G>T	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 Border Collie)	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

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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
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 (Discovered in the Boston Terrier) Variant 2	SGCD	CAT>GG	0	AR	Clear

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Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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
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 IIIB (Discovered in the Schipperke)	NAGLU	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 Labrador Retriever) Variant 1	COL6A3	G>A	0	AR	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

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Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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)	HCRT2	G>A	0	AR	Clear
Narcolepsy (Discovered in the Labrador Retriever)	HCRT2	G>A	0	AR	Clear
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 Border Collie)	CLN5	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

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Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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
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
Phosphofructokinase 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

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Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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
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
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

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Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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
Rod-Cone Dysplasia 3	PDE6A	Deletion	0	AR	Clear
Sensorineural Deafness (Discovered in the Rottweiler)	LOXHD1	G>C	0	AR	Clear
Sensory Neuropathy	FAM134B	Insertion	0	AR	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 (Discovered in the Alpine Dachsbracke)	SCN8A	G>T	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

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Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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
Trapped Neutrophil Syndrome	VPS13B	Deletion	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
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

Shadow

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Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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

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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.