Rosa

Breed: Boerboel
Microchip number: 900049000037498
Birth date: 2019-07-18

Registration number: 361740
Test date: 2021-03-20
ID kit: DNGRTRP

Rosa's Profile

Pet information

Registered name
Athletic Rose

Sex
F

Owner reported breed
Boerboel

Date of birth
2019-07-18

Microchip number
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Genetic Diversity

Rosa's Percentage of Heterozygosity
38%

Health summary

At Risk 0 conditions
Carrier 0 conditions
Clear 213 conditions

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Genetic Diversity

Heterozygosity

Rosa's Percentage of Heterozygosity

38%

Rosa's genome analysis shows an average level of genetic heterozygosity when compared with other Boerboels.

Typical Range for Boerboels

36% - 41%

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Health conditions known in the breed

Canine Multifocal Retinopathy 1

Gene	Risk Variant	Copies	Inheritance	Result
BEST1	C>T	0	AR	**Clear**

Information about the genetic condition

Typically, the first ocular fundus changes in CMR1 can be diagnosed by the age of four months. In many cases, the lesions may appear to heal or even go away, sometimes leaving no evidence or only a wrinkle at the site of the healed lesion. In almost all cases, lesions from CMR1 do not progress significantly over time, so there is generally no reduction in eyesight though more serious cases could exhibit vision impairment. Very seldom is the patient completely blinded.

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 CMR1 mutation can be safely bred with a clear dog with no copies of the CMR1 mutation. About half of the puppies will have one copy (carriers) and half will have no copies of the CMR1 mutation. A dog with two copies of the CMR1 mutation can be safely bred with a clear dog. The resulting puppies will be all carriers with one copy of the CMR1 mutation. Puppies in a litter which is 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 CMR1 mutation 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.

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Traits

Coat Color

	Gene	Variant	Copies	Result
Fawn Copies of this variant will cause dogs to show fawn if they do not have other variant that will mask this effect, such as a plain red, black or white coat.	ASIP	a^y	2	Fawn possible
Recessive Black	ASIP	a	0	No effect
Tan Points	ASIP	a^t	0	No effect
Dominant Black 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^B	1	Black or brindle possible
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	E^m	1	Dark Muzzle possible
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
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

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Color Modification

	Gene	Variant	Copies	Result
Red Intensity 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
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	1	No effect
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 (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

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Coat Patterns

	Gene	Variant	Copies	Result
Saddle Tan 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	-	2	Saddle possible

Coat Length and Curl

	Gene	Variant	Copies	Result
Long Hair (lh1)	FGF5	lh^{1}	0	No effect
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

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Shedding

	Gene	Variant	Copies	Result
Reduced Shedding One or two copies of the Reduced Shedding variant is likely to reduce a dog's tendency to shed. Copies of the Furnishings variant, particularly two, also reduce the tendency of a dog to shed.	MC5R	sd	2	Low shedder

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^{al}	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

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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

More Body Features

	Gene	Variant	Copies	Result
Back Muscle and Bulk One or two copies of the Back Muscle and Bulk variant may result in a bulkier more muscled back.	ACSL4	-	1	Bulky appearance likely
High Altitude Adaptation	EPAS1	-	0	No effect
Short Legs (Chondrodysplasia, CDPA)	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
Obesity risk (POMC)	POMC	Deletion	—	AD	Inconclusive
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 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
Canine Leukocyte Adhesion Deficiency (CLAD), type III	FERMT3	Insertion	0	AR	Clear
Canine Multifocal Retinopathy 2	BEST1	G>A	0	AR	Clear
Canine Multifocal Retinopathy 3	BEST1	Deletion	0	AR	Clear
Canine Scott Syndrome	ANO6	G>A	0	AR	Clear
Centronuclear Myopathy (Discovered in the Great Dane)	BIN1	A>G	0	AR	Clear
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

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Chondrodysplasia (Discovered in Norwegian Elkhound and Karelian Bear Dog)	ITGA10	C>T	0	AR	Clear
Cleft Lip & Palate with Syndactyly	ADAMTS20	Deletion	0	AR	Clear
Cleft Palate	DLX6	C>A	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
Congenital Dyshormonogenic Hypothyroidism with Goiter (Discovered in the Shih Tzu)	SLC5A5	G>A	0	AR	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 Myasthenic Syndrome (Discovered in the Golden Retriever)	COLQ	G>A	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

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Congenital Stationary Night Blindness (CSNB)	RPE65	A>T	0	AR	Clear
Craniomandibular Osteopathy (Discovered in Scottish Terrier breeds)	SLC37A2	C>T	0	AD	Clear
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
Deafness and Vestibular Dysfunction (DINGS1), (Discovered in Doberman Pinscher)	PTPRQ	Insertion	0	AR	Clear
Degenerative Myelopathy	SOD1	G>A	0	AR	Clear
Demyelinating Neuropathy	SBF2	G>T	0	AR	Clear
Dental Hypomineralization	FAM20C	C>T	0	AR	Clear
Dilated Cardiomyopathy (Discovered in the Schnauzer)	RBM20	Deletion	0	AR	Clear
Dominant Progressive Retinal Atrophy	RHO	C>G	0	AD	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 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
Early-Onset Progressive Retinal Atrophy (Discovered in the Portuguese Water Dog)	CCDC66	Insertion	0	AR	Clear
Epidermolytic Hyperkeratosis	KRT10	G>T	0	AR	Clear

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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
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
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
Goniodysgenesis and Glaucoma (Discovered in the Border Collie)	OLFML3	G>A	0	AR	Clear

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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 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 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
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
Intestinal Cobalamin Malabsorption (Discovered in the Beagle)	CUBN	Deletion	0	AR	Clear

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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
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
Lagotto Storage Disease	ATG4D	G>A	0	AR	Clear
Lamellar Ichthyosis	TGM1	Insertion	0	AR	Clear
Lethal Acrodermatitis (Discovered in the Bull Terrier)	MKLN1	A>C	0	AR	Clear
Ligneous Membranitis	PLG	T>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
MDR1 Medication Sensitivity	MDR1/ABCB1	Deletion	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

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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
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 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
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

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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 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
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
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
Polycystic Kidney Disease	PKD1	G>A	0	AD	Clear
Prekallikrein Deficiency	KLKB1	T>A	0	AR	Clear

Rosa

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Microchip number: 900049000037498
Birth date: 2019-07-18

Registration number: 361740
Test date: 2021-03-20
ID kit: DNGRTRP

Other health conditions tested

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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-PRA1 variant)	SLC4A3	Insertion	0	AR	Clear
Progressive Retinal Atrophy (Discovered in the Lhasa Apso)	IMPG2	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

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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
Rod-Cone Dysplasia 3	PDE6A	Deletion	0	AR	Clear
Sensory Ataxic Neuropathy	tRNATyr	Deletion	0	MT	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 (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

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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
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
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

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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.