

Roxanne

Breed: Labrador Retriever
Birth date: 2021-10-29
Owner: David P Cassens

Registry: American Kennel Club
Registration number: SS29881902
Test date: 2023-09-12
ID kit: DJNYMDT

Roxanne's Profile

Pet information

Registered name: Roxanne
Sex: F

Breed specific genetic health tests

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Alexander Disease	GFAP	G>A	0	AR	Clear
Centronuclear Myopathy (Discovered in the Labrador Retriever)	PTPLA	Insertion	0	AR	Clear
Congenital Cornification (Discovered in the Labrador Retriever)	NSDHL	Deletion	0	XD	Clear
Congenital Myasthenic Syndrome (Discovered in the Labrador Retriever)	COLQ	T>C	0	AR	Clear
Ehlers-Danlos Syndrome (Discovered in the Labrador Retriever)	COL5A1	Deletion	0	AD	Clear
Exercise-Induced Collapse	DNM1	G>T	0	AR	Clear
Hemophilia A (Discovered in the Labrador Retriever)	Confidential	-	0	XR	Clear
Hereditary Elliptocytosis	SPTB	C>T	0	AD	Clear
Hereditary Nasal Parakeratosis (Discovered in the Labrador Retriever)	SUV39H2	A>C	0	AR	Clear
Hyperuricosuria	SLC2A9	G>T	0	AR	Clear
Muscular Dystrophy-Dystroglycanopathy (Discovered in the Labrador Retriever)	LARGE	C>T	0	AR	Clear
Myotonia Congenita (Discovered in the Labrador Retriever)	CLCN1	T>A	0	AR	Clear
Narcolepsy (Discovered in the Labrador Retriever)	HCRT2	G>A	0	AR	Clear
Obesity risk (POMC)	POMC	Deletion	0	AD	Clear
Progressive Rod Cone Degeneration (prcd-PRA)	PRCD	G>A	0	AR	Clear

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Breed specific genetic health tests

Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Stargardt Disease (Discovered in the Labrador Retriever)	ABCA4	Insertion	0	AR	Clear
X-Linked Myotubular Myopathy	MTM1	C>A	0	XR	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.