

Registration number: FI10329/25 Test date: 2025-02-06 ID kit: DFQTMCJ



## Gigi's Profile

### Pet information

<b>Registered name</b> Chic Choix Tiny Jewels Mariko Ichiban	Sex F
<b>Owner reported breed</b> Lhasa Apso	<b>Date of birth</b> 2024-10-24
Microchip number 934000011367068	
Constia Diversity	

### **Genetic Diversity**

**Gigi's Percentage of Heterozygosity** 39%

### **Health summary**





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## Genetic Diversity

### Heterozygosity

### **Gigi's Percentage of Heterozygosity**

39%

Gigi's genome analysis shows an average level of genetic heterozygosity when compared with other Lhasa Apsos.

### Typical Range for Lhasa Apsos

29% - 39%

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## Health conditions known in the breed

Chondrodystrophy (CDDY) and Intervertebral Disc Disease (IVDD) Risk	Gene	Risk Variant	Copies	Inheritance	Result
	FGF4 retrogene	Insertion	0	AD	Clear

### ✤ Information about the genetic condition

Chondrodystrophy (CDDY) is a form of skeletal dysplasia which affects the development of cartilage and bone growth in a number of dog breeds. The associated CDDY genetic variant is an FGF4-retrogene insertion on dog chromosome 12, discovered by researchers in the Bannasch Laboratory at the University of California, Davis (Brown et al. 2017), and should not be confused with the FGF4-retrogene insertion on dog chromosome 18 (Parker et al. 2017), associated with a short-legged phenotype known as chondrodysplasia (CDPA). In dogs with CDDY, disproportionate growth (short limbs, normal sized body and head) can be observed as early as one week of age. CDDY follows a semi-dominant mode of inheritance. This means dogs with one copy of the genetic variant typically have some shortening of their legs, whereas dogs with two copies will show a more obvious shortening. Although not necessarily directly associated with CDDY, valgus limb deformities may be observed during physical examination of some dogs. However, affected dogs are more likely to experience premature degeneration and calcification of the intervertebral discs, a process also known as intervertebral disc disease (IVDD). Dogs with IVDD secondary to this genetic variant have an increased risk of intervertebral disc herniation (IVDH), consistent with Hansen Type I. The risk of developing IVDH follows a dominant mode of inheritance, meaning only one copy of this variant is needed to consider a dog predisposed for disc herniation. Age of onset of disc herniation appears to vary considerably between breeds, with the median age of dogs presenting for surgery varying from 3 years to 10 years. However, please note this variant is a risk factor and some dogs with one, or even two copies, of this variant may not go on to show signs of disc disease. It is worth clarifying that if disc herniation does not occur dorsally, a dog may appear asymptomatic as the spinal cord is less likely to be compressed. Additionally, not all dogs affected by IVDD have the FGF4-retrogene insertion found on chromosome 12, indicating additional genetic causes remain to be discovered.

### 🗴 Breeder recommendation

This variant is considered a risk factor for Chondrodystrophy (CDDY) and Intervertebral Disc Disease (IVDD), and dogs with one or two copies of the variant are at increased risk. However not all dogs with one or two copies of this variant will show signs of disc disease. Use of dogs with one or two copies of the CDDY and IVDD variant should be critically considered, as there is a risk that the resulting litter will contain affected puppies. For example, if a dog with one copy of the CDDY and IVDD variant is bred with a clear dog with no copies of the CDDY and IVDD variant, about half of the puppies will have one copy and half will have no copies of the CDDY and IVDD variant. Some breeds carry the variant at such a high rate that breeding dogs with one copy of the disorder is unavoidable. In such cases, mate selection should be planned to slowly reduce the frequency of the variant within the breed over time if possible. In breeds where both FGF4 retrogenes are present and a short stature is desirable, breeders can select for dogs positive for the CDPA (chromosome 18) variant, and against dogs with the CDDY (chromosome 12) variant to maintain breed-specific leg length. Please note: It is possible that clinical signs similar to the ones associated with the CDDY and IVDD variant could develop due to a different genetic or clinical cause.

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## Health conditions known in the breed

Hemophilia B (Discovered in the Lhasa Apso)	Gene	Risk Variant	Copies	Inheritance	Result
	FIX	Deletion	0	XR	Clear

#### ↔ Information about the genetic condition

Blood coagulation is a complex process. Factor IX is one of the proteins necessary for blood coagulation and its deficiency causes hemophilia B in an affected dog. Hemophilia B is a milder blood disorder than hemophilia A, but it may still cause life-threatening bleeding. Specific factor assay may be measured by a reference laboratory. Prior to surgery or invasive procedures, a prothrombin (PT) and partial thromboplastin time (PTT) should be measured. Additional supportive measures, including transfusions, may be necessary. Excessive and prolonged bleeding may be observed during tooth extractions, routine surgeries, and even minor traumas. The condition is usually more severe for active dogs of large size.

#### S Breeder recommendation

This disorder is X-linked recessive, meaning the genetic variant is found on the X chromosome. Given males only have one X chromosome, a single affected copy will increase the risk of being diagnosed with the disorder. Females typically require two copies to be at an elevated risk. Use of dogs with one or two copies of the variant is not recommended for breeding as there is a risk that the resulting litter will contain affected puppies. Please note: It is possible that clinical signs similar to the ones caused by this variant could develop due to a different genetic or clinical cause.

Progressive Retinal Atrophy (Discovered in the Lhasa Apso)	Gene	Risk Variant	Copies	Inheritance	Result
	IMPG2	Insertion	0	AR	Clear

#### ↔ Information about the genetic condition

In the Lhasa Apso breed, the onset of clinical signs is observed with initial loss of vision in dim light (night blindness) that gradually progresses to total blindness. The first signs are caused by loss of rod photoreceptor cells required for vision in dim light followed by reduction of the visual field and blindness as the cone cells of the retina also degenerate. The onset of clinical signs typically occurs at midage, although age of onset can vary.

#### 🗴 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 PRA mutation can be safely bred with a clear dog with no copies of the PRA mutation. About half of the puppies will have one copy (carriers) and half will have no copies of the PRA 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 PRA 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> 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	ау	2	Fawn possible
Recessive Black	ASIP	а	0	No effect
Tan Points	ASIP	at	0	No effect
Dominant Black	CBD103	K <sup>B</sup>	0	No effect
Mask	MC1R	Em	0	No effect
Recessive Red (e1)	MC1R	e1	0	No effect
Recessive Red (e2)	MC1R	e <sup>2</sup>	0	No effect
Recessive Red (e3)	MC1R	e <sup>3</sup>	0	No effect
Sable (Discovered in the Cocker Spaniel)	MC1R	е <sup>н</sup>	0	No effect
Widow's Peak (Discovered in Ancient dogs)	MC1R	eA	0	No effect
Widow's Peak (Discovered in the Afghan Hound and Saluki)	MC1R	Ec	0	No effect

### **Color Modification**

	Gene	Variant	Copies	Result
Cocoa (Discovered in the French Bulldog)	HPS3	CO	0	No effect
Red Intensity	MFSD12	i	1	No effect
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.				
Dilution (d1) Linkage test	MLPH	d1	0	No effect

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### **Color Modification**

	Gene	Variant	Copies	Result
Dilution (d2)	MLPH	d <sup>2</sup>	0	No effect
Dilution (d3)	MLPH	d³	0	No effect
Chocolate (basd)	TYRP1	basd	0	No effect
Chocolate (bc)	TYRP1	bc	0	No effect
Chocolate (bd)	TYRP1	bd	0	No effect
Chocolate (be)	TYRP1	be	0	No effect
Chocolate (bh)	TYRP1	bh	0	No effect
Chocolate (bs)	TYRP1	b₅	0	No effect

### **Coat Patterns**

	Gene	Variant	Copies	Result
<b>Piebald</b> Dog with copies of the Piebald variant are likely to show white spotting, patches and/or a white coat, with two copies having a greater effect than one, although the strength of this effect may be influenced by other genes.	MITF	Sp	1	White markings possible
Merle	PMEL	Μ	0	No effect
Harlequin	PSMB7	Н	0	No effect
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
Roan (Linkage test)	USH2A	Tr	0	No effect

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### **Coat Length and Curl**

	Gene	Variant	Copies	Result
Long Hair (lh1)	FGF5	lh1	2	Long coat
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 (Ih1) or two of any combination of long hair variants. However, there are other variants suspected to influence coat length.				
Long Hair (lh2)	FGF5	lh <sup>2</sup>	0	No effect
Long Hair (lh3)	FGF5	lh³	0	No effect
Long Hair (lh4)	FGF5	lh4	0	No effect
Long Hair (lh5)	FGF5	lh⁵	0	No effect
Curly Coat	KRT71	С	0	No effect

### Hairlessness

	Gene	Variant	Copies	Result
Hairlessness (Discovered in the Chinese Crested Dog) Linkage test	FOXI3	Hrcc	0	No effect
Hairlessness (Discovered in the American Hairless Terrier)	SGK3	hraht	0	No effect
Hairlessness (Discovered in the Scottish Deerhound)	SKG3	hrsd	0	No effect

### Shedding

	Gene	Variant	Copies	Result
Reduced Shedding	MC5R	sd	2	Low shedder
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.				

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### **More Coat Traits**

	Gene	Variant	Copies	Result
Hair Ridge	FGF3, FGF4, FGF19, ORAOV1	R	0	No effect
Furnishings	RSPO2	F	1	Furnishings likely
Dogs with one or two copies of the Furnishing variant are likely to display a fuzzy beard, moustache and eyebrows, but a long or curly coat will make this variant less apparent.				
Albino	SLC45A2	Cal	0	No effect

### **Head Shape**

	Gene	Variant	Copies	Result
Short Snout (BMP3 variant)	BMP3	-	0	No effect
Short Snout (SMOC2 variant)	SMOC2	-	1	Shortened snout likely
Copies of this skull shape variant usually results in a shorter snout, whereas dogs with no copies of this variant tend to have a longer snout.				

### Eye Color

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

### Ears

	Gene	Variant	Copies	Result
<b>Floppy Ears</b> Dogs with zero copies of this variant are more likely to have permanently upright or prick ears, and fully folded ears are more likely with two copies inherited. Please note however that many genetic variants influence ear carriage. Dogs with some cartilage stiffness to their ears can sometimes raise their ears upright when 'at alert' but will flop down when relaxed.	MSRB3	-	1	Partially floppy 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	1	Hind dewclaws possible
One or two copies of this Hind Dewclaws variant may result in your dog having hind dewclaws. Around half of the dogs				

in your dog having hind dewclaws. Around half of the dogs with one copy of this variant will have hind dewclaws, and it is possible for the dewclaws to be just on one leg. With two copies the trait is more likely to be expressed and could be more pronounced.

### **More Body Features**

	Gene	Variant	Copies	Result
Back Muscle and Bulk	ACSL4	-	0	No effect
High Altitude Adaptation	EPAS1	-	0	No effect
<b>Short Legs (Chondrodysplasia, CDPA)</b> Dogs with one copy of the Short Legs (CDPA) variant typically have some shortening of their legs, whereas with two copies there is more obvious shortening.	FGF4	-	2	Shortened legs likely
Short Legs (Chondrodystrophy, CDDY)	FGF4	-	0	No effect
Short Tail	T-box	Т	0	Full tail length likely

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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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Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Centronuclear Myopathy (Discovered in the Labrador Retriever)	PTPLA	Insertion	0	AR	Clear
Cerebellar Ataxia	RAB24	A>C	0	AR	Clear
Cerebellar Cortical Degeneration	SNX14	C>T	0	AR	Clear
Cerebellar Hypoplasia	VLDLR	Deletion	0	AR	Clear
Cerebral Dysfunction	SLC6A3	G>A	0	AR	Clear
Chondrodysplasia (Discovered in Norwegian Elkhound and Karelian Bear Dog)	ITGA10	C>T	0	AR	Clear
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
Congenital Cornification (Discovered in the Labrador Retriever)	NSDHL	Deletion	0	XD	Clear

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Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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)	СНАТ	G>A	0	AR	Clear
Congenital Stationary Night Blindness (CSNB)	RPE65	A>T	0	AR	Clear
Craniomandibular Osteopathy (Discovered in Scottish Terrier breeds)	SLC37A2	C>T	0	AD	Clear
Craniomandibular Osteopathy (Discovered in the Australian Terrier)	COL1A1	C>T	0	AD	Clear
Craniomandibular Osteopathy (Discovered in the Basset Hound)	SLC37A2	C>T	0	AD	Clear
Craniomandibular Osteopathy (Discovered in the Weimaraner)	SLC35D1	Deletion	0	AD	Clear
Cystic Renal Dysplasia and Hepatic Fibrosis	INPP5E	G>A	0	AR	Clear

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Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
Cystinuria Type I-A	SLC3A1	C>T	0	AR	Clear
Cystinuria Type II-A	SLC3A1	Deletion	0	AD	Clear
Darier Disease (Discovered in the Irish Terrier)	ATP2A2	Insertion	0	AD	Clear
Deafness and Vestibular Dysfunction (DINGS1), (Discovered in Doberman Pinscher)	PTPRQ	Insertion	0	AR	Clear
Deafness and Vestibular Dysfunction (DINGS2), (Discovered in Doberman Pinscher)	MYO7A	G>A	0	AR	Clear
Degenerative Myelopathy	SOD1	G>A	0	AR	Clear
Demyelinating Neuropathy	SBF2	G>T	0	AR	Clear
Dental 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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Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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
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

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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 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 Illa, (GSD Illa)	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
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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Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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)	SGCD	Deletion	0	AR	Clear
Limb-girdle Muscular Dystrophy, Type L3 (Discovered in the Miniature Dachshund)	SGCA	G>A	0	AR	Clear

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

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Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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
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
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

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Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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
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

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Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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 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

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Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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 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
Spongy Degeneration with Cerebellar Ataxia (Discovered in Belgian Malinois - SDCA1)	KCNJ10	T>C	0	AR	Clear
Spongy Degeneration with Cerebellar Ataxia (Discovered in Belgian Malinois - SDCA2)	ATP1B2	Insertion	0	AR	Clear
Stargardt Disease (Discovered in the Labrador Retriever)	ABCA4	Insertion	0	AR	Clear
Startle Disease (Discovered in Irish Wolfhounds)	SLC6A5	G>T	0	AR	Clear
Startle Disease (Discovered in the Miniature American Shepherd)	Confidential	-	0	AR	Clear

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Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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
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



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Genetic Condition	Gene	Risk Variant	Copies	Inheritance	Result
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.