



8700 3967 9218

Helmivuoren Dioriitti, Australian Shepherd

**Registered Name:** Helmivuoren Dioriitti

**Owner:** Sari Kariluoto

**Nickname:** Papu

**Country:** Finland

**Registration ID:** ER25916/16

**Testing date:** 2019/10/28

**Microchip:** 981098106337309

**Breed:** Australian Shepherd

**Gender:** Female

Dog's identity verified from microchip or tattoo by veterinarian or other authorised person during sample taking: **Yes**

## Test results - Known disorders in the breed

Disorder	Type	Mode of Inheritance	Result
Canine Multifocal Retinopathy 1, (CMR1); mutation originally found in Mastiff-related breeds	Ocular Disorders	Autosomal Recessive	Clear
Cone Degeneration, (CD) or Achromatopsia; mutation originally found in Alaskan Malamute	Ocular Disorders	Autosomal Recessive	Clear
Degenerative Myelopathy, (DM; SOD1A)	Neurological Disorders	Autosomal Recessive (Incomplete Penetrance)	Clear
Hyperuricosuria, (HUU)	Renal Disorders	Autosomal Recessive	Clear
Primary Hereditary Cataract, (PHC); mutation originally found in Australian Shepherd	Ocular Disorders	Autosomal Dominant (Incomplete Penetrance)	Clear

On behalf of Genoscoper Laboratories,

SIGNATURE

Jonas Donner, PhD, Head of Research and Development  
at Genoscoper Laboratories



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## Additional Tests Available to Purchase

Disorder	Type	Mode of Inheritance	Result
OptiGen® Collie Eye Anomaly, (CEA)	Ocular Disorders	Autosomal Recessive	Clear
OptiGen® Progressive Rod Cone Degeneration, (prcd-PRA)	Ocular Disorders	Autosomal Recessive	Clear

## Test results - New potential disorders in the breed

Disorder	Type	Mode of Inheritance	Result
Craniomandibular Osteopathy, (CMO); mutation associated with terrier breeds	Skeletal Disorders	Autosomal Dominant (Incomplete Penetrance)	 At risk (heterozygote)
Von Willebrand's Disease (vWD) Type 1	Blood Disorders	Autosomal Recessive	Clear

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## Test results for pharmacogenetics

Disorder	Mode of Inheritance	Result
Multi-Drug Resistance 1, (MDR1)	Autosomal Dominant	Clear

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## Test results - Traits - page 1

### Coat Type

Trait	Genotype	Description
Coat Length	I/I	The dog is genetically long-haired.
Furnishings / Improper Coat in Portuguese Water Dogs (marker test)	GG/CC	The dog is not genetically likely to express furnishings.
<i>KRT71</i> c.451C>T (p.Arg151Trp)	C/C	The dog does not carry any copies of the tested allele causing curly coat. The dog most likely has non-curly hair.
<i>MC5R</i> c.237A>T	C/C	The dog does not carry the tested allele associated with low shedding. This genotype has no effect on a dog with furnishings, but non-wire-haired dog with this genotype is likely heavy or seasonal shedder.
<i>SGK3</i> (p.Val96Glyfs)	I/I	The dog does not carry the tested hairlessness allele of the American Hairless Terrier.

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## Test results - Traits - page 2

### Coat Colour

Trait	Genotype	Description
Colour Locus E - Extensions	E/E	The dog is likely to express the coat colour defined by the K and A loci.
Colour Locus B - Brown	b/b    bb/B	Dog with this genotype may express brown or black pigment. The dog has black pigment, if the b alleles causing brown colour are located on the same chromosome.
Colour Locus K - Dominant Black	ky/ky	The dog is likely to express the coat colour defined by the colour locus A.
Colour Locus A - Agouti	at/a	The dog has genetically tan points or saddle tan pattern. The dog carries recessive black.
Colour Locus S - Piebald or extreme white spotting	S/S	The dog is likely to have solid coat colour with minimal white.
Colour Locus H - Harlequin	h/h	The dog doesn't have harlequin pattern.
Merle (M allele)	m/m	The dog is genetically non-merle and does not carry a S/LV gene SINE insertion.
Saddle Tan (RALY gene dupl.)	-/dup	The dog may have saddle tan pattern if it has also tan point genotype at the A locus.
Dilution (d <sup>2</sup> allele)	G/G	The dog does not carry any copies of the rare d <sup>2</sup> allele associated with dilution in Chow Chow, Sloughi and Thai Ridgeback.
Albinism (caL-allele)	C/C	The dog does not carry the tested mutation for albinism.

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## Test results - Traits - page 3

### Morphology

Trait	Genotype	Description
LIMBR1 DC-1	G/G	The dog does not carry the tested allele associated with hind dewclaws in Asian breeds. The dog is not likely to have hind dewclaws.
LIMBR1 DC-2	A/G	The dog carries one copy of the allele associated with hind dewclaws in western breeds. About 50% of the dogs with this genotype have hind dewclaws.
BMP3 c.1344C>A (p.Phe448Leu)	C/C	The dog does not carry the tested allele typically associated with shortened head (brachycephaly). The dog is more likely to have an elongated head (dolichocephaly).
chr10:11072007	T/T	The dog does not carry an allele typically associated with floppy ears. The dog is more likely to have pricked than floppy ears.
T c.189C>G (p.Ile63Met)	C/C	The dog does not carry the tested bobtail-causing genetic variant. The dog is most likely long-tailed.
EPAS1 (p.Gly305Ser)	G/G	The dog does not carry the tested variant associated with adaptation to high altitudes.

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## Test results - Traits - page 4

### Body Size

Trait	Genotype	Description
<i>IGF1</i> (chr15:41221438)	A/G	The dog is heterozygous for the ancestral allele. This means that it carries one copy of the genetic allele typically associated with small body mass and one copy typically associated with large body mass.
<i>IGF1R</i> c.611G>A (p.Arg204His)	G/G	The dog carries two ancestral alleles typically found in larger-sized breeds.
<i>FGF4</i> insertion	D/D	The dog is homozygous for the ancient allele. The dog is likely to have legs of normal length.
<i>STC2</i> (chr4:39182836)	A/T	The dog carries one copy of the allele associated with reduced body size and one copy of the allele associated with no size reduction.
<i>GHR1</i> (p.Glu191Lys)	G/G	The dog has two copies of the ancestral allele associated with larger body size.
<i>GHR2</i> (p.Pro177Leu)	C/C	The dog has two copies of the ancestral allele associated with larger body size.
<i>HMGA2</i> (chr10:8348804)	G/G	The dog has two copies of the ancestral allele associated with larger body size.

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## Test results - Additional disorders found in other breeds - page 1

### Blood Disorders - page 1

Disorder	Mode of Inheritance	Result
Bleeding disorder due to P2RY12 defect	Autosomal Recessive	Clear
Canine Cyclic Neutropenia, Cyclic Hematopoiesis, Grey Collie Syndrome, (CN)	Autosomal Recessive	Clear
Canine Leukocyte Adhesion Deficiency (CLAD), type III	Autosomal Recessive	Clear
Canine Scott Syndrome, (CSS)	Autosomal Recessive	Clear
Factor IX Deficiency or Hemophilia B; mutation Gly379Glu	X-linked Recessive	Clear
Factor IX Deficiency or Hemophilia B; mutation originally found in Airedale Terrier	X-linked Recessive	Clear
Factor IX Deficiency or Hemophilia B; mutation originally found in Lhasa Apso	X-linked Recessive	Clear
Factor VII Deficiency	Autosomal Recessive	Clear
Factor VIII Deficiency or Hemophilia A; mutation originally found in Boxer	X-linked Recessive	Clear
Factor VIII Deficiency or Hemophilia A; mutation originally found in German Shepherd Dog	X-linked Recessive	Clear
Factor VIII Deficiency or Hemophilia A; mutation originally found in Old English Sheepdog	X-linked Recessive	Clear
Factor VIII Deficiency or Hemophilia A; p.Cys548Tyr mutation originally found in German Shepherd	X-linked Recessive	Clear
Factor XI Deficiency	Autosomal Dominant (Incomplete Penetrance)	Clear
Familial Congenital Methemoglobinemia; mutation originally found in Pomeranian	Autosomal Recessive	Clear
Glanzmann Thrombasthenia Type I, (GT); mutation originally found in Pyrenean Mountain Dog	Autosomal Recessive	Clear
Glanzmann Thrombasthenia Type I, (GT); mutation originally found in mixed breed dogs	Autosomal Recessive	Clear
Hereditary Elliptocytosis		Clear
Hereditary Phosphofructokinase (PFK) Deficiency	Autosomal Recessive	Clear
Macrothrombocytopenia; disease-linked variant originally found in Norfolk and Caim Terrier	Autosomal Recessive	Clear
May-Hegglin Anomaly (MHA)	Autosomal Dominant	Clear
Prekallikrein Deficiency	Autosomal Recessive	Clear





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## Test results - Additional disorders found in other breeds - page 2

### Blood Disorders - page 2

Disorder	Mode of Inheritance	Result
Pyruvate Kinase Deficiency; mutation originally found in Basenji	Autosomal Recessive	Clear
Pyruvate Kinase Deficiency; mutation originally found in Beagle	Autosomal Recessive	Clear
Pyruvate Kinase Deficiency; mutation originally found in Pug	Autosomal Recessive	Clear
Pyruvate Kinase Deficiency; mutation originally found in West Highland White Terrier	Autosomal Recessive	Clear
Thrombopathia; mutation originally found in Basset Hound	Autosomal Recessive	Clear
Thrombopathia; mutation originally found in Eskimo Spitz	Autosomal Recessive	Clear
Thrombopathia; mutation originally found in Landseer	Autosomal Recessive	Clear
Trapped Neutrophil Syndrome, (TNS)	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 1	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 2	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 3; mutation originally found in Kooikerhondje	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 3; mutation originally found in Scottish Terrier	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 3; mutation originally found in Shetland Sheepdog	Autosomal Recessive	Clear



## Test results - Additional disorders found in other breeds - page 3

### Ocular Disorders - page 1

Disorder	Mode of Inheritance	Result
Canine Multifocal Retinopathy 2, (CMR2); mutation originally found in Coton de Tulear	Autosomal Recessive	Clear
Canine Multifocal Retinopathy 3, (CMR3); mutation originally found in Lapponian Herder	Autosomal Recessive	Clear
Cone Degeneration, (CD) or Achromatopsia; mutation originally found in German Shepherd Dog	Autosomal Recessive	Clear
Cone Degeneration, (CD) or Achromatopsia; mutation originally found in German Shorthaired Pointer	Autosomal Recessive	Clear
Cone-Rod Dystrophy 1, (crd1); mutation originally found in American Staffordshire Terrier	Autosomal Recessive	Clear
Cone-Rod Dystrophy 2, (crd2); mutation originally found in American Pit Bull Terrier	Autosomal Recessive	Clear
Cone-Rod Dystrophy, (cord1-PRA / crd4)	Autosomal Recessive (Incomplete Penetrance)	Clear
Cone-Rod Dystrophy, Standard Wirehaired Dachshund, (crd SWD)	Autosomal Recessive	Clear
Congenital Eye Disease; mutation originally found in Irish Soft-Coated Wheaten Terrier	Autosomal Recessive	Clear
Congenital Stationary Night Blindness, (CSNB)	Autosomal Recessive	Clear
Dominant Progressive Retinal Atrophy, (DPRA)	Autosomal Dominant	Clear
Early Retinal Degeneration, (erd); mutation originally found in Norwegian Elkhound	Autosomal Recessive	Clear
Generalized Progressive Retinal Atrophy	Autosomal Recessive	Clear
Golden Retriever Progressive Retinal Atrophy 1, (GR_PRA 1)	Autosomal Recessive	Clear
Golden Retriever Progressive Retinal Atrophy 2, (GR_PRA 2)	Autosomal Recessive	Clear
Primary Lens Luxation, (PLL)	Autosomal Recessive	Clear
Primary Open Angle Glaucoma, (POAG); mutation originally found in Basset Fauve de Bretagne	Autosomal Recessive	Clear
Primary Open Angle Glaucoma, (POAG); mutation originally found in Beagle	Autosomal Recessive	Clear
Primary Open Angle Glaucoma, (POAG); mutation originally found in Norwegian Elkhound	Autosomal Recessive	Clear
Primary Open Angle Glaucoma, (POAG); mutation originally found in Petit Basset Griffon Vendeen	Autosomal Recessive	Clear
Primary lens luxation (PLL) and glaucoma; mutation originally found in Shar Pei	Autosomal Recessive	Clear



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## Test results - Additional disorders found in other breeds - page 4

### Ocular Disorders - page 2

Disorder	Mode of Inheritance	Result
Progressive Retinal Atrophy (PRA4); mutation originally found in Lhasa Apso	Autosomal Recessive	Clear
Progressive Retinal Atrophy Type III, (PRA type III); mutation originally found in Tibetan Spaniel and Tibetan Terrier	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (CNGA1-PRA); mutation originally found in Shetland Sheepdog	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (PAP1_PRA); mutation originally found in Papillon and Phalene	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (PRA); mutation originally found in Basenji	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (PRA); mutation originally found in Swedish Vallhund	Autosomal Recessive	Clear
Rod-Cone Dysplasia 1, (rcd1); mutation originally found in Irish Setter	Autosomal Recessive	Clear
Rod-Cone Dysplasia 1a, (rcd1a); mutation originally found in Sloughi	Autosomal Recessive	Clear
Rod-Cone Dysplasia 3, (rcd3)	Autosomal Recessive	Clear
X-Linked Progressive Retinal Atrophy 1, (XL PRA1)	X-linked Recessive	Clear
X-Linked Progressive Retinal Atrophy 2, (XL PRA2; Type A PRA)	X-linked Recessive	Clear

### Cardiac Disorders

Disorder	Mode of Inheritance	Result
Dilated Cardiomyopathy, (DCM); mutation originally found in Schnauzer	Autosomal Recessive	Clear
Long QT Syndrome	Autosomal Dominant	Clear

### Endocrine Disorders

Disorder	Mode of Inheritance	Result
Congenital Hypothyroidism; mutation originally found in Tenterfield Terrier	Autosomal Recessive	Clear
Congenital Hypothyroidism; mutation originally found in Toy Fox and Rat Terrier	Autosomal Recessive	Clear



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## Test results - Additional disorders found in other breeds - page 5

### Immunological Disorders

Disorder	Mode of Inheritance	Result
Autosomal Recessive Severe Combined Immunodeficiency, (ARSCID)	Autosomal Recessive	Clear
Complement 3 (C3) Deficiency	Autosomal Recessive	Clear
Myeloperoxidase Deficiency	Autosomal Recessive	Clear
Severe Combined Immunodeficiency in Frisian Water Dogs, (SCID)	Autosomal Recessive	Clear
X-Linked Severe Combined Immunodeficiency (XSCID); mutation originally found in Basset Hound	X-linked Recessive	Clear
X-Linked Severe Combined Immunodeficiency (XSCID); mutation originally found in Cardigan Welsh Corgi	X-linked Recessive	Clear



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## Test results - Additional disorders found in other breeds - page 6

### Renal Disorders

Disorder	Mode of Inheritance	Result
2,8-Dihydroxyadenine (2,8-DHA) urolithiasis	Autosomal Recessive	Clear
Cystinuria Type I-A; mutation originally found in Newfoundland Dog	Autosomal Recessive	Clear
Cystinuria Type II-A; mutation originally found in Australian Cattle Dog	Autosomal Dominant	Clear
Familial Nephropathy (FN); mutation originally found in English Cocker Spaniel	Autosomal Recessive	Clear
Familial Nephropathy (FN); mutation originally found in English Springer Spaniel	Autosomal Recessive	Clear
Fanconi Syndrome	Autosomal Recessive	Clear
Polycystic Kidney Disease in Bull Terriers, (BTPKD)	Autosomal Dominant	Clear
Primary Hyperoxaluria, (PH); mutation originally found in Coton de Tulear	Autosomal Recessive	Clear
Protein Losing Nephropathy, (PLN); NPHS1 gene variant		Clear
Renal Cystadenocarcinoma and Nodular Dermatofibrosis, (RCND)	Autosomal Dominant	Clear
X-Linked Hereditary Nephropathy, (XLHN)	X-linked Recessive	Clear
X-Linked Hereditary Nephropathy, (XLHN); mutation originally found in Navasota Dog	X-linked Recessive	Clear
Xanthinuria, Type 1a; mutation originally found in mixed breed dogs	Autosomal Recessive	Clear
Xanthinuria, Type 2a; mutation originally found in Toy Manchester Terrier	Autosomal Recessive	Clear
Xanthinuria, Type 2b; mutation originally found in Cavalier King Charles Spaniel and English Cocker Spaniel	Autosomal Recessive	Clear



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## Test results - Additional disorders found in other breeds - page 7

### Metabolic Disorders

Disorder	Mode of Inheritance	Result
Glycogen Storage Disease Type II or Pompe's Disease, (GSD II)	Autosomal Recessive	Clear
Glycogen Storage Disease Type IIIa, (GSD IIIa)	Autosomal Recessive	Clear
Glycogen Storage Disease Type Ia, (GSD Ia)	Autosomal Recessive	Clear
Hypocatalasia or Acatlasemia	Autosomal Recessive	Clear
Intestinal Cobalamin Malabsorption or Imerslund-Gräsbeck Syndrome, (IGS); mutation originally found in Beagle	Autosomal Recessive	Clear
Intestinal Cobalamin Malabsorption or Imerslund-Gräsbeck Syndrome, (IGS); mutation originally found in Border Collie	Autosomal Recessive	Clear
Mucopolysaccharidosis Type IIIA, (MPS IIIA); mutation originally found in Dachshund	Autosomal Recessive	Clear
Mucopolysaccharidosis Type IIIA, (MPS IIIA); mutation originally found in New Zealand Huntaway	Autosomal Recessive	Clear
Mucopolysaccharidosis Type VII, (MPS VII); mutation originally found in Brazilian Terrier	Autosomal Recessive	Clear
Mucopolysaccharidosis Type VII, (MPS VII); mutation originally found in German Shepherd	Autosomal Recessive	Clear
Pyruvate Dehydrogenase Phosphatase 1 (PDP1) Deficiency	Autosomal Recessive	Clear



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## Test results - Additional disorders found in other breeds - page 8

### Muscular Disorders

Disorder	Mode of Inheritance	Result
Cavalier King Charles Spaniel Muscular Dystrophy, (CKCS-MD)	X-linked Recessive	Clear
Centronuclear Myopathy, (CNM); mutation originally found in Great Dane	Autosomal Recessive	Clear
Centronuclear Myopathy, (CNM); mutation originally found in Labrador Retriever	Autosomal Recessive	Clear
Duchenne or Dystrophin Muscular Dystrophy, (DMD); mutation originally found in Golden Retriever	X-linked Recessive	Clear
Duchenne or Dystrophin Muscular Dystrophy, (DMD); mutation originally found in Norfolk Terrier	X-linked Recessive	Clear
Muscular Dystrophy, Ullrich-type; mutation originally found in Landseer	Autosomal Recessive	Clear
Myostatin deficiency (Double Muscling, "Bully")	Autosomal Recessive	Clear
Myotonia Congenita; mutation originally found in Australian Cattle Dog	Autosomal Recessive	Clear
Myotonia Congenita; mutation originally found in in Labrador Retriever	Autosomal Recessive	Clear
Myotubular Myopathy; mutation originally found in Rottweiler	X-linked Recessive	Clear
Nemaline Myopathy; mutation originally found in American Bulldog	Autosomal Recessive	Clear
X-Linked Myotubular Myopathy	X-linked Recessive	Clear



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## Test results - Additional disorders found in other breeds - page 9

### Neurological Disorders - page 1

Disorder	Mode of Inheritance	Result
Acral Mutilation Syndrome, (AMS)	Autosomal Recessive	Clear
Alaskan Husky Encephalopathy, (AHE)	Autosomal Recessive	Clear
Alexander Disease (AxD); mutation originally found in Labrador Retriever	Autosomal Dominant	Clear
Bandera's Neonatal Ataxia, (BNAt)	Autosomal Recessive	Clear
Benign Familial Juvenile Epilepsy or Remitting Focal Epilepsy	Autosomal Recessive	Clear
Cerebellar Cortical Degeneration, (CCD); mutation originally found in Vizsla	Autosomal Recessive	Clear
Cerebral Dysfunction; mutation originally found in Friesian Stabyhoun	Autosomal Recessive	Clear
Dandy-Walker-Like Malformation (DWLM); mutation originally found in Eurasier	Autosomal Recessive	Clear
Early-Onset Progressive Polyneuropathy; mutation originally found in Alaskan Malamute	Autosomal Recessive	Clear
Early-Onset Progressive Polyneuropathy; mutation originally found in Greyhound	Autosomal Recessive	Clear
Fetal Onset Neuroaxonal Dystrophy, (FNAD)	Autosomal Recessive	Clear
Hereditary Ataxia or Cerebellar Ataxia; mutation originally found in Old English Sheepdog and Gordon Setter	Autosomal Recessive	Clear
Hereditary Ataxia; mutation originally found in in Norwegian Buhund	Autosomal Recessive	Clear
Hyperekplexia or Startle Disease	Autosomal Recessive	Clear
Hypomyelination; mutation originally found in Weimaraner	Autosomal Recessive	Clear
Juvenile Myoclonic Epilepsy, (JME); mutation originally found in Rhodesian Ridgeback	Autosomal Recessive	Clear
Juvenile encephalopathy; mutation originally found in Parson Russell Terrier	Autosomal Recessive	Clear
L-2-Hydroxyglutaric aciduria, (L2HGA); mutation originally found in Staffordshire Bull Terrier	Autosomal Recessive	Clear
L-2-Hydroxyglutaric aciduria, (L2HGA); mutation originally found in West Highland White Terrier	Autosomal Recessive	Clear
Lagotto Storage Disease, (LSD)	Autosomal Recessive	Clear
Neonatal Cerebellar Cortical Degeneration or Cerebellar Abiotrophy, (NCCD)	Autosomal Recessive	Clear





## Test results - Additional disorders found in other breeds - page 10

### Neurological Disorders - page 2

Disorder	Mode of Inheritance	Result
Neonatal Encephalopathy with Seizures, (NEWS)	Autosomal Recessive	Clear
Neuroaxonal Dystrophy (NAD); mutation originally found in Spanish Water Dog	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 1, (NCL1); mutation originally found in Dachshund	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 10, (NCL10); mutation originally found in American Bulldog	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 4A, (NCL4); mutation originally found in American Staffordshire Terrier	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 5, (NCL5); mutation originally found in Border Collie	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 8, (NCL8); mutation originally found in Alpine Dachsbracke	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 8, (NCL8); mutation originally found in Australian Shepherd	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 8, (NCL8); mutation originally found in English Setter	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis, (NCL7); mutation originally found in Chinese Crested Dog and Chihuahua	Autosomal Recessive	Clear
Polyneuropathy with ocular abnormalities and neuronal vacuolation, (POANV); mutation originally found in Black Russian Terrier	Autosomal Recessive	Clear
Progressive Early-Onset Cerebellar Ataxia; mutation originally found in Finnish Hound	Autosomal Recessive	Clear
Sensory Neuropathy; mutation originally found in Border Collie	Autosomal Recessive	Clear
Shaking Puppy Spongiform LeucoEncephaloMyelopathy, (SLEM); mutation originally found in Border Terrier	Autosomal Recessive	Clear
Spinocerebellar Ataxia with Myokymia and/or Seizures (SCA)	Autosomal Recessive	Clear
Spinocerebellar Ataxia/ Late-Onset Ataxia (SCA, LOA)	Autosomal Recessive	Clear
Spongy Degeneration with Cerebellar Ataxia, (SDCA1); mutation originally found in Belgian Shepherd Dog	Autosomal Recessive	Clear
Spongy Degeneration with Cerebellar Ataxia, (SDCA2); mutation originally found in Belgian Shepherd Dog	Autosomal Recessive	Clear
X-Linked Tremors; mutation originally found in English Springer Spaniel	X-linked Recessive	Clear



## Test results - Additional disorders found in other breeds - page 11

### Neuromuscular Disorders

Disorder	Mode of Inheritance	Result
Congenital Myasthenic Syndrome (CMS); mutation originally found in Labrador Retriever	Autosomal Recessive	Clear
Congenital Myasthenic Syndrome, (CMS); mutation originally found in Jack Russell Terrier	Autosomal Recessive	Clear
Congenital Myasthenic Syndrome, (CMS); mutation originally found in Old Danish Pointing Dog	Autosomal Recessive	Clear
Exercise-Induced Collapse, (EIC)	Autosomal Recessive (Incomplete Penetrance)	Clear
GM1 Gangliosidosis; mutation originally found in Alaskan Husky	Autosomal Recessive	Clear
GM1 Gangliosidosis; mutation originally found in Portuguese Water Dog	Autosomal Recessive	Clear
GM1 Gangliosidosis; mutation originally found in Shiba Dog	Autosomal Recessive	Clear
GM2 Gangliosidosis, mutation originally found in Japanese Chin	Autosomal Recessive	Clear
GM2 Gangliosidosis; mutation originally found in Toy Poodle	Autosomal Recessive	Clear
Globoid Cell Leukodystrophy or Krabbe Disease, (GLD); mutation originally found in Irish Setter	Autosomal Recessive	Clear
Globoid Cell Leukodystrophy or Krabbe Disease, (GLD); mutation originally found in Terriers	Autosomal Recessive	Clear
Paroxysmal Dyskinesia, (PxD); mutation originally found in Irish Soft Coated Wheaten Terrier	Autosomal Recessive	Clear



## Test results - Additional disorders found in other breeds - page 12

### Skeletal Disorders

Disorder	Mode of Inheritance	Result
Chondrodysplasia; mutation originally found in Norwegian Elkhound and Karelian Bear Dog	Autosomal Recessive	Clear
Cleft Palate; Cleft Lip and Palate with Syndactyly; ADAMTS20 gene mutation originally found in Nova Scotia Duck Tolling Retriever	Autosomal Recessive	Clear
Cleft Palate; DLX6 gene mutation originally found in Nova Scotia Duck Tolling Retriever	Autosomal Recessive	Clear
Hereditary Vitamin D-Resistant Rickets, (HVDRR)	Autosomal Recessive	Clear
Oculoskeletal Dysplasia 2 or Dwarfism-Retinal Dysplasia 2, (OSD2)	Autosomal Recessive	Clear
Osteochondrodysplasia; mutation originally found in Miniature Poodle	Autosomal Recessive	Clear
Osteochondromatosis; mutation originally found in American Staffordshire Terrier	Autosomal Dominant	Clear
Osteogenesis Imperfecta, (OI); mutation originally found in Beagle	Autosomal Dominant	Clear
Osteogenesis Imperfecta, (OI); mutation originally found in Dachshund	Autosomal Recessive	Clear
Skeletal Disease; mutation originally found in Karelian Bear Dog	Autosomal Recessive	Clear
Skeletal Dysplasia 2, (SD2)	Autosomal Recessive	Clear
Spondylocostal Dysostosis	Autosomal Recessive	Clear
Van den Ende-Gupta Syndrome, (VDEGS)	Autosomal Recessive	Clear



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## Test results - Additional disorders found in other breeds - page 13

### Dermal Disorders

Disorder	Mode of Inheritance	Result
Dystrophic Epidermolysis Bullosa; mutation originally found in Central Asian Ovcharka	Autosomal Recessive	Clear
Dystrophic Epidermolysis Bullosa; mutation originally found in Golden Retriever	Autosomal Recessive	Clear
Epidermolytic Hyperkeratosis	Autosomal Recessive	Clear
Focal Non-Epidermolytic Palmoplantar Keratoderma, (FNEPPK); mutation originally found in Dogue de Bordeaux	Autosomal Recessive	Clear
Golden Retriever Ichthyosis	Autosomal Recessive	Clear
Hereditary Footpad Hyperkeratosis, (HFH)	Autosomal Recessive	Clear
Hereditary Nasal Parakeratosis, (HNPK); mutation originally found in Greyhound	Autosomal Recessive	Clear
Ichthyosis; mutation originally found in American Bulldog	Autosomal Recessive	Clear
Ichthyosis; mutation originally found in Great Dane	Autosomal Recessive	Clear
Lamellar Ichthyosis, (LI)	Autosomal Recessive	Clear
Lethal Acrodermatitis, (LAD); mutation originally found in in Bull Terrier and Miniature Bull Terrier	Autosomal Recessive	Clear
Ligneous Membranitis	Autosomal Recessive	Clear
Musladin-Lueke syndrome, (MLS)	Autosomal Recessive	Clear
X-Linked Ectodermal Dysplasia, (XHED)	X-linked Recessive	Clear



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## Test results - Additional disorders found in other breeds - page 14

### Other Disorders

Disorder	Mode of Inheritance	Result
Acute Respiratory Distress Syndrome, (ARDS); mutation originally found in Dalmatian	Autosomal Recessive	Clear
Amelogenesis Imperfecta, (AI); mutation originally found in Italian Greyhound	Autosomal Recessive	Clear
Amelogenesis Imperfecta, (AI); mutation originally found in Parson Russell Terrier	Autosomal Recessive	Clear
Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis, (CKCSID)	Autosomal Recessive	Clear
Dental Hypomineralisation; mutation originally found in Border Collie	Autosomal Recessive	Clear
Narcolepsy; mutation originally found in Dachshund	Autosomal Recessive	Clear
Narcolepsy; mutation originally found in Doberman Pinscher	Autosomal Recessive	Clear
Narcolepsy; mutation originally found in Labrador Retriever	Autosomal Recessive	Clear
Persistent Müllerian Duct Syndrome, (PMDS); mutation originally found in Miniature Schnauzer	Autosomal Recessive	Clear
Primary Ciliary Dyskinesia, (PCD)	Autosomal Recessive	Clear



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## APPENDIX

### Explanation of the results of the tested disorders

#### Autosomal recessive inheritance (ARI)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

Carrier - A dog carries one copy of the tested mutation. Carriers typically have a normal, healthy appearance but pass on the mutation to approximately 50% of their offspring.

At risk - A dog carries two copies of the tested mutation and is at high or increased risk of developing the disease/condition.

#### Autosomal dominant inheritance (ADI)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

At risk - A dog carries one or two copies of the tested mutation and is at high or increased risk of developing the disease/condition.

#### X-linked recessive inheritance (X-linked)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

Carrier - Female carriers typically have a normal, healthy appearance but carry one copy of the tested mutation on one of their X chromosomes. As males only have one X chromosome, there are no male carriers.

At risk - Female dogs at risk carry two mutated copies of the tested mutation. Males carry one copy of the tested mutation on their single X chromosome. Dogs at risk are at high or increased risk of developing the disease/condition.

Please note that the descriptions above are generalized based on typically observed inheritance patterns. When obtaining a 'carrier' or 'at risk' test result, always refer to the corresponding online test documentation for more detailed information on the condition and any exceptions.

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