8700 4011 5475

Jamamba Very-Merry, Poodle - Standard (FCI size standard) - Grey, apricot and red

**Registered Name:** Jamamba Very-Merry

**Nickname:** Jamamba

**Registration ID:** EST-00756/16

**Microchip:** 900032001748753

**Breed:** Poodle - Standard (FCI size standard) - Grey, apricot and red

**Gender:** Female

**Owner:** Irina Seeder

**Country:** Estonia

**Testing date:** 2020/5/24

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 |
|---|---|---|---|
| Degenerative Myelopathy, (DM; SOD1A) | Neurological Disorders | Autosomal Recessive (Incomplete Penetrance) | Clear |
| Neonatal Encephalopathy with Seizures, (NEWS) | Neurological Disorders | Autosomal Recessive | Clear |
| Osteochondrodysplasia; mutation originally found in Miniature Poodle | Skeletal Disorders | Autosomal Recessive | Clear |
| Von Willebrand's Disease (vWD) Type 1 | Blood Disorders | Autosomal Recessive | Clear |

## Additional Tests Available to Purchase

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

On behalf of Genoscoper Laboratories,

SIGNATURE

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

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**Testing date:** 2020/5/24

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

## Test results for pharmacogenetics

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

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**Country:** Estonia

**Testing date:** 2020/5/24

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

## 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) | AA/TT | The dog is genetically likely to express furnishings. |
| *KRT71* c.451C>T (p.Arg151Trp) | T/T | The dog carries two copies of the tested allele causing curly coat. The dog is likely to have curly hair, if it is long-haired. |
| *MC5R* c.237A>T | C/T | The dog carries one copy of the allele associated with heavy shedding and one copy of the allele associated 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. |
| *SGK3* (p.Val96Gly*fs*) | I/I | The dog does not carry the tested hairlessness allele of the American Hairless Terrier. |

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**Gender:** Female

**Owner:** Irina Seeder
**Country:** Estonia
**Testing date:** 2020/5/24

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

## Test results - Traits - page 2

### Coat Colour

| Trait | Genotype | Description |
|---|---|---|
| Colour Locus E - Extensions | e/e | The dog has recessive red coat colour. |
| Colour Locus B - Brown | B/B | The dog is not likely to have brown pigment. |
| Colour Locus K - Dominant Black | KB/KB | The dog is genetically dominant black. |
| Colour Locus A - Agouti | at/at | The dog has genetically tan points or saddle tan pattern. |
| 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. |
| Dilution ($d^2$ allele) | D/D | The dog does not carry any copies of the rare d2 allele associated with dilution in Chow Chow, French Bulldog, Sloughi and Thai Ridgeback. |
| Merle (M allele) | m/m | The dog is genetically non-merle and does not carry a *SILV* 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. |
| Albinism ($c^{aL}$-allele) | C/C | The dog does not carry the tested mutation for albinism. |

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**Gender:** Female

**Owner:** Irina Seeder
**Country:** Estonia
**Testing date:** 2020/5/24

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

## Test results - Traits - page 3

### Body Size

| Trait | Genotype | Description |
|---|---|---|
| *IGF1* (chr15:41221438) | G/G | The dog is homozygous for the ancestral allele typically associated with large body mass. |
| *IGF1R* c.611G>A (p.Arg204His) | G/G | The dog carries two ancestral alleles typically found in larger-sized breeds. |
| *ACSL4* chrX.82919525C>T | C/C | The dog doesn't have the allele associated with large skeletal size and heavy muscling with considerable back fat thickness. |
| *IGSF1* p.Asp768Glu | A/C | The dog has one copy of the allele associated with heavy muscling. |
| *IRS4* chrX:82296039 | G/G | The dog doesn't have the allele associated with large body size. |
| *FGF4* insertion | D/D | The dog is homozygous for the ancient allele. The dog is likely to have legs of normal length. |
| *STC2* (chr4:39182836) | T/T | The dog has two copies of the ancestral allele associated with larger body size. |
| *GHR1* (p.Glu191Lys) | G/G | The dog has two copies of the ancestral allele associated with larger body size. |
| *GHR2* (p.Pro177Leu) | C/C | The dog has two copies of the ancestral allele associated with larger body size. |
| *HMGA2* (chr10:8348804) | G/G | The dog has two copies of the ancestral allele associated with larger body size. |

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**Testing date:** 2020/5/24

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

## Test results - Traits - page 4

### Morphology

| Trait | Genotype | Description |
|---|---|---|
| *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). |
| *SMOC2* | D/D | 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 | C/T | The dog carries one copy of an allele typically associated with floppy ears, and one copy of an allele typically associated with pricked 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. |
| *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 | G/G | The dog does not carry the tested allele associated with hind dewclaws in western breeds. The dog is likely not to have hind dewclaws. |
| *AXL4* | D/D | The dog does not have the tested allele typically associated with blue eyes in Siberian Huskies. The dog is likely to have brown eyes. |

On behalf of Genoscoper Laboratories,

Jonas Donner

SIGNATURE

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

# 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 Cairn Terrier | Autosomal Recessive | Clear |
| May-Hegglin Anomaly (MHA) | Autosomal Dominant | Clear |
| Prekallikrein Deficiency | Autosomal Recessive | Clear |

# 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 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 1, (CMR1); mutation originally found in Mastiff-related breeds | Autosomal Recessive | Clear |
| 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 Alaskan Malamute | 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 |
| 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 Hereditary Cataract, (PHC); mutation originally found in Australian Shepherd | Autosomal Dominant (Incomplete Penetrance) | 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 |

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

## Ocular Disorders - page 2

| Disorder | Mode of Inheritance | Result |
|---|---|---|
| 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 |
| 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, (rdc1a); mutation originally found in Sloughi | Autosomal Recessive | Clear |
| Rod-Cone Dysplasia 3, (rcd3) | Autosomal Recessive | Clear |
| X-Linked Progressive Retinal Atrophy 1, (XLPRA1) | X-linked Recessive | Clear |
| X-Linked Progressive Retinal Atrophy 2, (XLPRA2; 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 |

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

### 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 |

### 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 |

## 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 |
| Hyperuricosuria, (HUU) | 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 |

## 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 Acatalasemia | 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 |

## 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 |

# 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 |
|---|---|---|
| Neuroaxonal Dystrophy (NAD); mutation originally found in Spanish Water Dog | Autosomal Recessive | Clear |
| Neuroaxonal Dystrophy, (NAD); mutation originally found in Papillon | 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 |
| Craniomandibular Osteopathy, (CMO); mutation associated with terrier breeds | Autosomal Dominant (Incomplete Penetrance) | Clear |
| Hereditary Vitamin D-Resistant Rickets, (HVDRR) | Autosomal Recessive | Clear |
| Oculoskeletal Dysplasia 2 or Dwarfism-Retinal Dysplasia 2, (OSD2) | 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 (Hypophosphatasia); 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 |

## 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 |

## 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 Dermatosis, (CKCSID) | Autosomal Recessive | Clear |
| Dental Hypomineralisation; mutation originally found in Border Collie | Autosomal Recessive | Clear |
| Lung Developmental Disease; mutation originally found in in Airedale Terrier | 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 |

# 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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