



8700 3319 6367

Keano dello Spirito del Bosco, Lagotto Romagnolo - Romagna Water Dog

**Registered Name:** Keano dello Spirito del Bosco

**Owner:** Estermann Irene

**Nickname:** Keano

**Country:** Switzerland

**Registration ID:** 751973

**Testing date:** 2018/5/19

**Microchip:** 756097200098984

**Breed:** Lagotto Romagnolo - Romagna Water Dog

**Gender:** Male

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 |
|---|------------------------|---------------------|--------|
| Benign Familial Juvenile Epilepsy or Remitting Focal Epilepsy | Neurological Disorders | Autosomal Recessive | Clear  |
| Hyperuricosuria, (HUU)  | Renal Disorders        | Autosomal Recessive | Clear  |
| Lagotto Storage Disease, (LSD)                                | Neurological Disorders | Autosomal Recessive | Clear  |

## Test results - New potential disorders in the breed

| Disorder  | Type                   | Mode of Inheritance | Result |
|---|------------------------|---------------------|--------|
| Neuroaxonal Dystrophy (NAD); mutation originally found in Spanish Water Dog | Neurological Disorders | Autosomal Recessive | Clear  |

On behalf of Genoscooper Laboratories,

SIGNATURE

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



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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) | 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. |
| SGK3   | 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      | The dog is likely to have brown pigment.   |
| Colour Locus K - Dominant Black                    | KB/KB    | The dog is genetically dominant black.   |
| Colour Locus A - Agouti                            |          |  |
| Colour Locus S - Piebald or extreme white spotting | sp/sp    | The dog is likely to have piebald spotting or to be extreme white.               |
| Colour Locus H - Harlequin                         | h/h      | The dog doesn't have harlequin pattern.  |
| Albinism (caL-allele)                              | C/C      | This dog does not carry the tested mutation for albinism.                        |
| Merle (M allele)                                   | m/m      | This dog is genetically non-merle and does not carry a SILV gene SINE insertion. |
| Saddle Tan (RALY gene dupl.)                       | dup/dup  | The dog may have tan points if it has tan point genotype at the A locus.         |

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## Test results - Traits - page 3

### Morphology

| Trait                               | Genotype | Description   |
|-------------------------------------|----------|---|
| <i>BMP3</i> 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). |
| <i>T</i> 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.  |
| chr10:11072007                      | C/C      | The dog carries two copies of an allele typically associated with floppy ears. The dog is more likely to have floppy than pricked ears.                               |
| <i>EPAS1</i> (p.Gly305Ser)          | G/G      | The dog does not carry the tested variant associated with adaptation to high altitudes.   |
| <i>LIMBR1</i> 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.                                  |
| <i>LIMBR1</i> 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.                                |

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## Test results - Traits - page 4

### Body Size

| Trait                               | Genotype | Description  |
|-------------------------------------|----------|--|
| <i>IGF1</i> (chr15:41221438)        | A/A      | The dog is homozygous for the derived allele typically associated with small 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)         | T/T      | The dog has two copies of the ancestral allele associated with larger body size.               |
| <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 German Wirehaired Pointer     | X-linked Recessive                         | Clear   |
| Factor IX Deficiency or Hemophilia B; mutation originally found in Lhasa Apso                    | X-linked Recessive                         | No call |
| 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   |
| 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   |



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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 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  |
| Congenital Stationary Night Blindness, (CSNB)   | Autosomal Recessive                         | Clear  |
| Dominant Progressive Retinal Atrophy, (DPRA)  | Autosomal Dominant                          | Clear  |
| Generalized Progressive Retinal Atrophy   | Autosomal Recessive                         | Clear  |
| Golden Retriever Progressive Retinal Atrophy 1, (GR_PRA 1)  | 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  |
| Primary Open Angle Glaucoma, (POAG); mutation originally found in Petit Basset Griffon Vendéen    | Autosomal Recessive                         | Clear  |



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

### Ocular Disorders - page 2

| Disorder   | Mode of Inheritance | Result |
|--|---------------------|--------|
| Primary lens luxation (PLL) and glaucoma; mutation originally found in Shar Pei  | 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, (XLPRA1)   | X-linked Recessive  | Clear  |
| X-Linked Progressive Retinal Atrophy 2, (XLPRA2)   | 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 |
|---|---------------------|--------|
| 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  |
| Cystinuria, Type II-B; mutation originally found in Miniature Pinscher                                      | Autosomal Dominant  | 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  |



## 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 Miniature Schnauzer                            | 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  |
| 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  |
| 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  |
| 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  |
| Neonatal Cerebellar Cortical Degeneration or Cerebellar Abiotrophy, (NCCD)                                  | Autosomal Recessive | Clear  |
| 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  |



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

### Neurological Disorders - page 2

| Disorder   | Mode of Inheritance | Result |
|--|---------------------|--------|
| 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  |
| Spinal Dysraphism  | 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  |
| X-Linked Tremors; mutation originally found in English Springer Spaniel  | X-linked Recessive  | Clear  |



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## 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  |
| 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  |
| 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  |
| Hereditary Footpad Hyperkeratosis, (HFH)   | 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  |
| Ligneous Membranitis   | Autosomal Recessive | Clear  |
| Musladin-Lueke syndrome, (MLS)   | Autosomal Recessive | Clear  |
| X-Linked Ectodermal Dysplasia, (XHED)  | X-linked Recessive  | Clear  |

### Pharmacogenetics

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