



8700 2369 1526

Ch. Nostryffelns Mozzafiata Mimi, Lagotto Romagnolo - Romagna Water Dog

**Registered Name:** Ch. Nostryffelns Mozzafiata Mimi

**Owner:** Janelle Schramko

**Nickname:** Mimi

**Country:** Australia

**Registration ID:** SE48453/2011

**Testing date:** 2016/11/18

**Microchip:** 752098100587739

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

**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 |
|---|----------------------|---------------------|--------|
| Benign Familial Juvenile Epilepsy or Remitting Focal Epilepsy | Neurologic Disorders | Autosomal Recessive | Clear  |
| Hyperuricosuria, (HUU)  | Renal Disorders      | Autosomal Recessive | Clear  |
| Lagotto Storage Disease, (LSD)                                | Neurologic 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 | Neurologic Disorders | Autosomal Recessive | Clear  |
| Von Willebrand's Disease (WVD) Type II                                      | Blood 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.   |
| Curly coat   | T/T      | The dog has a curly appearance and it carries two copies of the genetic variant typically associated with a curly coat. |

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**Gender:** Female

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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                              | bc/bs                      | The dog is likely to have brown coat.  |
| Colour Locus K - Dominant Black                     | KB/KB    KB/kbr    kbr/kbr | The dog is genetically dominant black or brindle.  |
| 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  | 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.  |
| Colour Locus C - Albinism (ca <sup>L</sup> -allele) | C/C                        | This dog does not carry the tested mutation for albinism.                                  |
| Colour Pattern (RALY gene): Saddle Tan              | dup/dup                    | The dog may have tan points if it has tan point genotype at the A locus.                   |

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**Gender:** Female

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

## Test results - Traits - page 3

### Morphology

| Trait   | Genotype | Description  |
|---|----------|--|
| Tiny size, insulin-like growth factor 1 receptor (IGF1R) gene variant                                       | G/G      | Your dog is homozygous for a genetic variant typically found in larger-sized breeds (height at the withers > 25.4 cm (10 inches)).   |
| Ear erectness (pricked ears versus floppy ears), variant chr10:11072007                                     | C/C      | Your dog is homozygous for (carries two copies of) a genetic variant typically associated with floppy ears. This genotype is common in breeds like English Springer Spaniel, Leonberger, Saluki, and Dachshunds. Interestingly, the C-allele of this variant is the ancestral allele frequent in wolf. |
| Bobtail   | C/C      | The dog does not carry any copy of the bobtail mutation. It therefore likely has a long-tailed phenotype.  |
| Snout/skull length (shortened head versus elongated head), bone morphogenetic protein 3 (BMP3) gene variant | C/C      | Your dog is homozygous for the genetic variant typically found in breeds with an elongated head (e.g. Saluki, Collie, Irish Wolfhound).  |
| Body mass, insulin-like growth factor 1 (IGF1) gene variant   | A/A      | The dog is homozygous for the genetic variant typically associated with small body mass. This genotype is common e.g. in Yorkshire Terrier, Chihuahua and Chinese Crested Dogs.  |

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

### Blood Disorders

| 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 (4 mutations)  | X-linked Recessive  | Clear  |
| Factor VII Deficiency   | Autosomal Recessive | Clear  |
| Factor VIII Deficiency or Hemophilia A (3 mutations)  | X-linked Recessive  | Clear  |
| Factor XI Deficiency  | Autosomal Recessive | Clear  |
| Glanzmann Thrombasthenia Type I, (GT); mutation originally found in Pyrenean Mountain Dog   | 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  |
| Pyruvate Kinase Deficiency (4 mutations)  | Autosomal Recessive | Clear  |
| Thrombopathia (3 mutations)   | Autosomal Recessive | Clear  |
| Trapped Neutrophil Syndrome, (TNS)  | Autosomal Recessive | Clear  |



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

### Ocular Disorders - page 1

| Disorder   | Mode of Inheritance                         | Result |
|--|---|--------|
| Canine Multifocal Retinopathy 1, (CMR1); Mastiff-related breeds mutation   | 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 (3 mutations)   | 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 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  |
| 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 Beagle   | Autosomal Recessive                         | Clear  |
| Primary Open Angle Glaucoma, (POAG); mutation originally found in Norwegian Elkhound                                   | 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  |



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

### Ocular Disorders - page 2

| Disorder  | Mode of Inheritance | Result |
|---|---------------------|--------|
| Rod-Cone Dysplasia 1, (rcd1) and Rod-Cone Dysplasia 1a, (rcd1a) (2 mutations) | 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 |
|------------------|---------------------|--------|
| Long QT Syndrome | Autosomal Dominant  | Clear  |

### Endocrine Disorders

| Disorder                                | Mode of Inheritance | Result |
|---|---------------------|--------|
| Congenital Hypothyroidism (2 mutations) | Autosomal Recessive | Clear  |

### Immunologic Disorders

| Disorder  | Mode of Inheritance | Result |
|---|---------------------|--------|
| Autosomal Recessive Severe Combined Immunodeficiency, (ARSCID)  | Autosomal Recessive | Clear  |
| Complement 3 (C3) Deficiency                                    | Autosomal Recessive | Clear  |
| Myeloperoxidase Deficiency                                      |                     | Clear  |
| Severe Combined Immunodeficiency in Frisian Water Dogs, (SCID)  | Autosomal Recessive | Clear  |
| X-linked Severe Combined Immunodeficiency (XSCID) (2 mutations) | X-linked Recessive  | Clear  |



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

### 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) (2 mutations)                     | X-linked Recessive  | Clear  |

### 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) (2 mutations) | Autosomal Recessive | Clear  |
| Mucopolysaccharidosis Type IIIA, (MPS IIIA) (2 mutations)                              | Autosomal Recessive | Clear  |
| Mucopolysaccharidosis Type VII, (MPS VII) (2 mutations)                                | Autosomal Recessive | Clear  |
| Pyruvate Dehydrogenase Phosphatase 1 (PDP1) Deficiency                                 | Autosomal Recessive | Clear  |





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

### Muscular Disorders

| Disorder  | Mode of Inheritance | Result |
|---|---------------------|--------|
| Cavalier King Charles Spaniel Muscular Dystrophy, (CKCS-MD)                                     | X-linked Recessive  | Clear  |
| Centronuclear Myopathy, (CNM) (2 mutations)   | 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 (MDL), Ullrich-type; mutation originally found in Landseer                   | Autosomal Recessive | Clear  |
| Muscular Hypertrophy (Double Muscling)  | Autosomal Recessive | Clear  |
| Myotonia Congenita (2 mutations)  | Autosomal Recessive | Clear  |
| X-Linked Myotubular Myopathy (2 mutations)  | X-linked Recessive  | Clear  |



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

### Neurologic Disorders - page 1

| Disorder  | Mode of Inheritance | Result |
|---|---------------------|--------|
| Alaskan Husky Encephalopathy, (AHE)   | Autosomal Recessive | Clear  |
| Bandera's Neonatal Ataxia, (BNAt)   | Autosomal Recessive | Clear  |
| Dandy-Walker-Like Malformation (DWLM); mutation originally found in Eurasier                                | Autosomal Recessive | Clear  |
| Cerebral Dysfunction; mutation originally found in Friesian Stabyhoun                                       | Autosomal Recessive | Clear  |
| Early-Onset Progressive Polyneuropathy (2 mutations)  | 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  |
| L-2-Hydroxyglutaric aciduria, (L2HGA); mutation originally found in Staffordshire Bull Terrier              | Autosomal Recessive | Clear  |
| Neonatal Cerebellar Cortical Degeneration or Cerebellar Abiotrophy, (NCCD)                                  | Autosomal Recessive | Clear  |
| Neonatal Encephalopathy with Seizures, (NEWS)   | 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 12, (NCL12)  | Autosomal Recessive | Clear  |
| Neuronal Ceroid Lipofuscinosis 8, (NCL8) (2 mutations)  | Autosomal Recessive | Clear  |
| Neuronal Ceroid Lipofuscinosis, (NCL7); mutation originally found in Chinese Crested Dog and Chihuahua      | Autosomal Recessive | Clear  |
| Progressive Early-Onset Cerebellar Ataxia; mutation originally found in Finnish Hound                       | 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  |



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

### Neurologic Disorders - page 2

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

### Neuromuscular Disorders

| Disorder  | Mode of Inheritance | Result |
|---|---------------------|--------|
| Congenital Myasthenic Syndrome, (CMS); mutation originally found in Jack Russell Terrier    | Autosomal Recessive | Clear  |
| Congenital Myasthenic Syndrome (CMS); mutation originally found in Labrador Retriever       | Autosomal Recessive | Clear  |
| Congenital Myasthenic Syndrome, (CMS); mutation originally found in Old Danish Pointing Dog | Autosomal Recessive | Clear  |
| GM1 Gangliosidosis (3 mutations)  | Autosomal Recessive | Clear  |
| GM2 Gangliosidosis or Sandhoff Disease (2 mutations)  | Autosomal Recessive | Clear  |
| Globoid Cell Leukodystrophy or Krabbe's Disease, (GLD) (2 mutations)                        | Autosomal Recessive | Clear  |



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

### 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; Cleft Lip and Palate with Syndactyly; 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  |
| Osteochondrodysplasia; mutation originally found in Miniature Poodle  | Autosomal Recessive                        | Clear  |
| Osteogenesis Imperfecta, (OI); mutation originally found in Beagle  |  | Clear  |
| Osteogenesis Imperfecta, (OI); mutation originally found in Dachshund   | 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 9

### Dermal Disorders

| Disorder   | Mode of Inheritance | Result |
|--|---------------------|--------|
| Dystrophic Epidermolysis Bullosa (2 mutations)               | Autosomal Recessive | Clear  |
| Epidermolytic Hyperkeratosis                                 | Autosomal Recessive | Clear  |
| Focal Non-Epidermolytic Palmoplantar Keratoderma (FNEPPK/DH) |                     | Clear  |
| Hereditary Footpad Hyperkeratosis, (HFH)                     | 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  |



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

### Other Disorders

| Disorder   | Mode of Inheritance | Result |
|--|---------------------|--------|
| 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 (3 mutations)   | 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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