2/12/2014

MyDogDNA PASS
8700 2321 7880 383
Come Se Avrei Zampe d'Orsa, Lagotto Romagnolo

**Registered name:** Come Se Avrei Zampe d'Orsa
**Nickname:** Zeta
**Registration ID:** ER18164/12
**Microchip:** 985170002363882
**Breed:** Lagotto Romagnolo
**Gender:** Female

**Owner:** Terhi Järvenpää
**Country:** Finland
**Testing date:** 14/2/2014
**DNA identification profile:** Identified with standard ISAG markers

Dog's identity verified from microchip or tattoo by veterinarian or other authorized 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 |
| Ivermectin sensitivity (MDR1) | Pharmacogenetics | Autosomal Recessive | Clear |
| Malignant Hyperthermia (MH) | Pharmacogenetics | Autosomal Dominant | Clear |

## Test results - New potential disorders in the breed

| Disorder | Type | Mode of inheritance | Result |
|---|---|---|---|
| Hyperuricosuria and Hyperuricemia (HUU) or Urolithiasis | Kidney disorders | Autosomal Recessive | Clear |

*When obtaining a carrier or affected test result, we recommend that you contact your veterinarian for more detailed information on the condition and possible treatment.*

*On behalf of Genoscoper Laboratories,*

Jonas Donner

SIGNATURE

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

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## Test results - Traits

| Trait | Genotype | Description |
|---|---|---|
| Colour Locus A | at/at | The dog is homozygous for at-allele. |
| Colour Locus B | bs/bs | Your dog is homozygous for bs allele. |
| Colour Locus E | e/E | The dog is heterozygous for e allele and E allele. |
| Colour Locus H | h/h | The dog is homozygous for h allele. |
| Colour Locus K | KB/KB \|\| KB/kbr \|\| kbr/kbr | The dog is homozygous for three nucleotide deletion. |
| Furnishings / Improper Coat in Portuguese Water Dogs (marker test) | AA/TT | The dog is genetically likely to express furnishings. |
| Body mass, insulin-like growth factor 1 (IGF1) gene variant | G/G | The dog is homozygous for the genetic variant typically associated with large body mass. This genotype is common e.g. in Great Dane, Newfoundland Dog and Greater Swiss Mountain Dog. |
| Coat length / "Fluffy" in Welsh Corgi | T/T | The dog carries two copies of the genetic variant typically associated with a long-haired coat. Dogs with this genotype typically have long coat. |
| 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. |
| 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. |
| Natural Bobtail (T-box mutation) | 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). |
| Tiny size, insulin-like growth factor 1 receptor (IGF1R) gene variant | A/G | Your dog is heterozygous for this variant. This means that your dog carries one copy of a genetic variant typically associated with tiny size (height at the withers < 25.4 cm (10 inches)), and one copy typically associated with larger size (> 25.4 cm (10 inches)). |

*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 1/6

### Blood disorders

| Disorder | Mode of inheritance | Result |
|---|---|---|
| Bleeding disorder due to P2RY12 defect | Autosomal Recessive | Clear |
| Canine Cyclic Neutropenia (Gray Collie Syndrome) | Autosomal Recessive | Clear |
| Canine Leukocyte Adhesion Deficiency (CLAD), type I | Autosomal Recessive | Clear |
| Factor IX Deficiency or Haemophilia B, Gly379Glu mutation | X-linked Recessive | Clear |
| Factor IX Deficiency or Haemophilia B; mutation originally found in Lhasa Apso | X-linked Recessive | Clear |
| Factor VII Deficiency | Autosomal Recessive | Clear |
| Factor VIII deficiency or Haemophilia A; mutation originally found in German Shepherd Dog | X-linked Recessive | Clear |
| Glanzmann Thrombasthenia (GT), Type I; mutation originally found in Pyrenean Mountain Dog | Autosomal Recessive | Clear |
| Glycogen Storage Disease VII or Hereditary Phosphofructokinase (PFK) Deficiency | Autosomal Recessive | Clear |
| May-Hegglin Anomaly (MHA) | Autosomal Dominant | Clear |
| Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Beagle | Autosomal Recessive | Clear |
| Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Labrador Retriever | Autosomal Recessive | Clear |
| Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Pug | Autosomal Recessive | Clear |
| Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier | Autosomal Recessive | Clear |
| Trapped Neutrophil Syndrome (TNS) | Autosomal Recessive | Clear |
| Von Willebrand's Disease (vWD) Type III; mutation originally found in Kooikerhondje | Autosomal Recessive | Clear |
| Von Willebrand's Disease (vWD) Type III; mutation originally found in Shetland Sheepdog | Autosomal Recessive | Clear |

### Cardiological disorders

| Disorder | Mode of inheritance | Result |
|---|---|---|
| Dilated Cardiomyopathy; mutation originally found in Doberman Pinscher (USA) | Autosomal Dominant | Clear |

## Test results - Additional disorders found in other breeds - page 2/6

### Endocrine disorders

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

### Eye disorders

| Disorder | Mode of inheritance | Result |
|---|---|---|
| Achromatopsia or Cone Degeneration (CD); mutation originally found in German Shorthaired Pointer | Autosomal Recessive | Clear |
| Autosomal Dominant Progressive Retinal Atrophy (ADPRA) | Autosomal Dominant | Clear |
| 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-rod Dystrophy (cord1-PRA / crd4) | Autosomal Recessive | Clear |
| Cone-rod dystrophy (crd SWD); mutation originally found in Standard Wire-haired Dachshund | Autosomal Recessive | Clear |
| Congenital Stationary Night Blindness (CSNB) | Autosomal Recessive | Clear |
| Generalized Progressive Retinal Atrophy; mutation originally found in Schapendoes | 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; mutation originally found in Beagle | 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 |

## Test results - Additional disorders found in other breeds - page 3/6

### Immunological disorders

| Disorder | Mode of inheritance | Result |
|---|---|---|
| ARSCID (Autosomal Recessive Severe Combined Immunodeficiency) | Autosomal Recessive | Clear |
| C3 deficiency | 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 |

### Kidney disorders

| Disorder | Mode of inheritance | Result |
|---|---|---|
| Autosomal Recessive Hereditary Nephropathy (ARHN); mutation originally found in English Cocker Spaniel | Autosomal Recessive | Clear |
| Autosomal Recessive Hereditary Nephropathy (ARHN); mutation originally found in English Springer Spaniel | Autosomal Recessive | Clear |
| Polycystic Kidney Disease (PKD) | Autosomal Dominant | Clear |
| Primary hyperoxaluria (PH); mutation originally found in Coton de Tulear | Autosomal Recessive | Clear |
| X-linked Hereditary Nephropathy (XLHN) | X-linked Recessive | Clear |

### Metabolic disorders

| Disorder | Mode of inheritance | Result |
|---|---|---|
| Glycogen Storage Disease, Type Ia (GSDIa) | Autosomal Recessive | Clear |
| Glycogen Storage Disease, type II or Pompe's disease | Autosomal Recessive | Clear |
| Glycogen Storage Disease, type IIIa (GSDIIIa) | Autosomal Recessive | Clear |
| Hypocatalasia or Acatalasemia | Autosomal Recessive | Clear |
| Mucopolysaccharidosis Type IIIA (MPSIIIA); mutation originally found in Dachshund | Autosomal Recessive | Clear |
| Mucopolysaccharidosis Type VI (MPSVI); mutation originally found in Poodle | Autosomal Recessive | Clear |
| Mucopolysaccharidosis Type VII (MPSVII); mutation originally found in Brazilian Terrier | Autosomal Recessive | Clear |
| Pyruvate Dehydrogenase Deficiency | Autosomal Recessive | Clear |

## Test results - Additional disorders found in other breeds - page 4/6

### Muscular disorders

| Disorder | Mode of inheritance | Result |
|---|---|---|
| Cavalier King Charles Spaniel Muscular Dystrophy (CKCS-MD) | X-linked Recessive | Clear |
| Duchenne-like Muscular Dystrophy, Pembroke Welsh Corgi-type | X-linked Recessive | Clear |
| Muscular Dystrophy, Duchenne type or Golden Retriever Muscular Dystrophy (GRMD) | X-linked Recessive | Clear |
| Myotonia; mutation originally found in Miniature Schnauzer | Autosomal Recessive | Clear |
| Myotubular Myopathy 1 or X-linked Myotubular Myopathy | X-linked Recessive | Clear |

### Neurological disorders

| Disorder | Mode of inheritance | Result |
|---|---|---|
| Cerebellar abiotrophy or neonatal cerebellar cortical degeneration (NCCD) | Autosomal Recessive | Clear |
| Fetal-onset Neuroaxonal Dystrophy (FNAD) | Autosomal Recessive | Clear |
| Hyperekplexia or Startle Disease | Autosomal Recessive | Clear |
| L-2-Hydroxyglutaric aciduria (L2HGA); mutation 1 originally found in Staffordshire Bull Terrier | Autosomal Recessive | Clear |
| L-2-Hydroxyglutaric aciduria (L2HGA); mutation 2 originally found in Staffordshire Bull Terrier | Autosomal Recessive | Clear |
| Neonatal Encephalopathy with Seizures (NEWS) | Autosomal Recessive | Clear |
| Neuronal Ceroid Lipofuscinosis 1 (NCL1) | Autosomal Recessive | Clear |
| Neuronal Ceroid Lipofuscinosis 10 (NCL10) | Autosomal Recessive | Clear |
| Neuronal Ceroid Lipofuscinosis 2 (NCL2) | Autosomal Recessive | Clear |
| Neuronal Ceroid Lipofuscinosis 5 (NCL5) | Autosomal Recessive | Clear |
| Neuronal Ceroid Lipofuscinosis 6 (NCL6) | Autosomal Recessive | Clear |
| Neuronal Ceroid Lipofuscinosis, type 12, mutation originally found in Tibetan terrier | Autosomal Recessive | Clear |
| Polyneuropathy; mutation originally found in Alaskan Malamute | Autosomal Recessive | Clear |
| Polyneuropathy; mutation originally found in Greyhound | Autosomal Recessive | Clear |
| Progressive early-onset cerebellar ataxia; mutation originally found in Finnish Hound | Autosomal Recessive | Clear |

## Test results - Additional disorders found in other breeds - page 5/6

### Neuromuscular disorders

| Disorder | Mode of inheritance | Result |
|---|---|---|
| Alpha Fucosidosis | Autosomal Recessive | Clear |
| Episodic falling (EF) | 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 Toy Poodle | Autosomal Recessive | Clear |
| Globoid Cell Leukodystrophy (GLD) or Krabbe's disease, Terrier mutation | Autosomal Recessive | Clear |

### Skeletal disorders

| Disorder | Mode of inheritance | Result |
|---|---|---|
| Chondrodysplasia (dwarfism); mutation originally found in Norwegian Elkhound and Karelian Bear Dog | Autosomal Recessive | Clear |
| Craniomandibular Osteopathy (CMO) | Autosomal Dominant | Clear |
| Osteogenesis imperfecta (OI) or Brittle Bone Disease; mutation originally found in Dachshund | Autosomal Recessive | Clear |
| Skeletal Dysplasia 2 (SD2) | Autosomal Recessive | Clear |

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

### Skin disorders

| Disorder | Mode of inheritance | Result |
|---|---|---|
| Ectodermal dysplasia or Skin Fragility Syndrome (ED-SFS) | Autosomal Recessive | Clear |
| Epidermolysis bullosa, dystrophic | Autosomal Recessive | Clear |
| Epidermolytic Hyperkeratosis or Ichthyosis in Norfolk Terrier | Autosomal Recessive | Clear |
| Musladin-Lueke syndrome (MLS) | Autosomal Recessive | Clear |

### Other disorders

| Disorder | Mode of inheritance | Result |
|---|---|---|
| Congenital Keratoconjuctivitis Sicca and Ichthyosiform Dermatosis (CKCSID) or Dry Eye Curly Coat Syndrome | Autosomal Recessive | Clear |
| Gallbladder Mucocele Formation | Autosomal Dominant | Clear |
| Narcolepsy; mutation originally found in Dobermann | Autosomal Recessive | Clear |
| Persistant Mullerian Duct Syndrome (PMDS), mutation originally found in Miniature Schnauzer | Autosomal Recessive | Clear |
| Primary Ciliary Dyskinesia (PCD) | Autosomal Recessive | Clear |

*On behalf of Genoscoper Laboratories,*

Jonas Donner

SIGNATURE

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

## 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.
*Affected* - 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.
*Affected* - 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.
*Affected* - Affected female dogs carry two mutated copies of the tested mutation. Affected males carry one copy of the tested mutation on their single X chromosome. Affected dogs 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 affected test result, always refer to the corresponding online test documentation for more detailed information on the condition and any exceptions.

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